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Dr. Raimundo Llanio Navarro

Dr. Aracelys Lantigua Cruz Dr.
Mercedes Batule Batule Dr. Miguel
Matarama Peñate
Dr. Enrique Arús Soler Dr.
Anayda Fernández Naranjo Dr.
Manuel Paniagua Estévez Dr.
Enrique Galbán García Dr.
Elsa García Bacallao

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Ing. Virgilia Salcines Batista

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National Information Center for Medical Sciences Calle I No. 202 corner a Línea, 11th floor, El Vedado,

City of Havana, 10400, Cuba

Email: ecimed @ infomed.sld.cu

Telephones: 55-3375 and 832 5338

Dr. Raimundo Llanio Navarro.

Doctor in Medical Sciences. Professor of Merit at the University of Havana. Full Professor of Clinical Propedeutics and Internal Medicine. Full Member of the Academy of Sciences. Second Degree Specialist in Gastroenterology. President of the Cuban Society of Gastroenterology and Endoscopy. Director of the Institute of Gastroenterology. Senior Researcher. Member of the Society of Internal Medicine.

Dr. Aracelys Lantigua Cruz.

Doctor in Medical Sciences. Full Professor of Medical Genetics. Second Degree Specialist in Medical Genetics. Head of the Department of Clinical Genetics of the Center for Medical Genetics.

Dr. Mercedes Batule Batule.

Doctor in Medical Sciences. Professor of Clinical Propedeutics and Internal Medicine. Second Degree Specialist in Internal Medicine. Member of the Rector's Advisory Council. Member of the Society of Internal Medicine.

Dr. Miguel Matarama Peñate.

Full Professor of Clinical Propedeutics and Internal Medicine. Second Degree Specialist in Internal Medicine. Head of the Clinical Department of the Calixto García Faculty. Member of the Cuban Society of Internal Medicine and Gastroenterology.

Dr. Enrique Arús Soler.

Doctor in Medical Sciences. Full Professor of the Higher Institute of Medical Sciences of Havana. Second Degree Specialist in Gastroenterology. Auxiliary Researcher: Head of the Gastroenterology Service of the " Hermanos Ameijeiras" Hospital. Vice President of the Cuban Society of Gastroenterology and Endoscopy.

Dr. Anayda Fernández Naranjo.

Full Professor of the Higher Institute of Medical Sciences of Havana. Second Degree Specialist in Human Anatomy. Dean of the “Julio Trigo” Faculty of Medical Sciences. City of Havana

Dr. Manuel Paniagua Estévez.

Doctor in Medical Sciences. Full Professor of Propedeutics and Internal Medicine. Second Degree Specialist in Gastroenterology. First Degree Specialist in Internal Medicine. Assistant Researcher. Teaching Assistant Director of the Institute of Gastroenterology. Member and Secretary of the Cuban Society of Gastroenterology and Endoscopy.

Dr. Enrique Galbán García

Assistant Professor of Epidemiology. Second Degree Specialist in Epidemiology. Assistant Researcher. Epidemiologist at the Institute of Gastroenterology.

Dr. Elsa García Bacallao

Second Degree Specialist in Gastroenterology.

Instructor Professor of Clinical Propedeutics.

Member of the Cuban Society of Gastroenterology and Digestive Endoscopy.

PROLOGUE

In this book that we present to doctors, who have so little time to update themselves and who suffer the tormenting onslaught of the information industry with the controversies that it causes, we intend to provide them in summary form with a large part of the syndromes that make up medicine, so that they can quickly recognize them if they are faced with patients suffering from them.

It is an incontrovertible fact that what is unknown cannot be diagnosed and our effort in writing this book makes it possible for the doctor to periodically update himself in this important field.

The structure, always the same, consists of: name, synonymy, syndromography (clinical and paraclinical examinations), syndromogenesis and etiology and bibliography.

We have not included evolution, prognosis or treatment, since these are sometimes multiple and variable since they depend on the different etiologies of the syndrome and are constantly subject to modifications due to the development of technology and science.

In each syndrome, the essential symptomatology for diagnosis is collected and is also complemented with specific aspects of syndromogenesis and etiology, which over the years will provide new elements, especially in some genetic syndromes or in others in which their etiology is currently unknown.

As for the bibliography at the end of each syndrome, we have tried to put the original and one or more revisions. In addition, there is a general bibliography of the complex works that we have resorted to for research on the topics, in which the bibliography corresponding to the syndromes that lack it will be found at the end of their description.

We have made a great effort to group the syndromes by specialties, although many share symptoms from several disciplines, however, they have been placed taking into account the main symptoms for which the patient consults. We hope that this type of grouping will be useful to the reader, which is not the general trend in the few books on syndromes written, and that they are always placed in alphabetical order, but we estimate that it will be easier for different specialists to find the syndromes that are part of each specialty.

We consider that we have fulfilled the fundamental objective that we set when we conceived this work, which does not attempt to enumerate, much less develop, the multiple syndromes of medicine. If we had done so, not only would it be a work in several volumes, but it would not fulfill the desire and hope that we put into its methodology and writing.

We never had the opportunity during our studies and medical exercises to have a work of this type. That is why Syndromes has been written with the intention that it will serve, in their daily work, medical students and general practitioners, although we are sure that it will be very useful to internists and specialists in different branches.

There is a saying that says: "the worst enemy of the good is the perfect" and this old phrase contains a lot of truth and is the reason why works of a certain category do not yield the usefulness that their authors expected. This explains why we have not elaborated, except on some syndromes, to try to complete the data presented; since doing so would have lost the true meaning of our intention in writing this work, as synthetic and practical as possible, especially when the level of those for whom it was conceived is analyzed. I hope for your understanding.

We thought it might be useful to list, at the end of each of the specialties or parts that make up the book, the syndromes that share common elements (many of which are described here), 1which will undoubtedly contribute to diagnostic thinking in the clinic.

Also included was a glossary of the terms less frequently used in medicine and a genetic glossary that are intended to make it easier for the reader to define the term, so that they do not have to look for a specialized dictionary, which is not always at hand.

Finally, the alphabetical index of syndromes has included not only the name of the syndrome, but an adequate number of synonyms by which it is indicated in the different texts.

We do not want to finish this presentation or prologue without recording our deep gratitude to the group of collaborators who helped us so much, they are: the co-authors; Clodomiro Alpízar designer, now deceased; Dulce María León, our editor; the secretaries Iraida Guma, Carmen Fleitas, Ana Gamoneda, Esperanza Macías, María Isabel Otero, who were in charge of the tedious work of typing the text. Estrella Morales responsible for adding the bibliography; the doctors: Miguel Soneiras, who reviewed the names and synonyms, and Castro Falcón, for his contribution to otorhinolaryngological syndromes, the colleagues of the Editorial Ciencias Médicas for their dedication and effort in the completion of this work; Silvia Gutiérrez for her advice and help as associate editor and especially to my lifelong companion Flora Villate for her collaboration in the related syndromes, the glossary, the constant corrections of this very complex work and above all for her patience and understanding during these 11 years that we have invested in compiling and producing the text.

If this reference book is of practical use for students and doctors, we will have the satisfaction that the effort made by all of us was worth it and that will be the best reward for the years invested in its preparation.

Prof. Raimundo Llanio Navarro

INTRODUCTION

Clinical medicine begins with the patient's symptoms and has the ultimate goal of detecting the disease that afflicts them; However, this process is not always easy and requires knowledge that the doctor must master to face the patient.

Years ago a small book was written entitled From Symptom to Disease, by Coste, and we would like to point out that it is essential to go through the syndrome before concluding this path. This is the logical sequence that must be taught to the student in the clinical rooms, and the diagnostic process necessary for this purpose requires good technical and theoretical learning.

However, in the clinic it is essential to classify the patient by the syndrome or syndromes they present in order to guide the diagnostic discussion and scientific thinking.

The use of the term syndrome has become so widespread that this concept is now sometimes being used for a group of problems that have or do not have to do with medicine, such as Vietnam syndrome, Gulf War syndrome, fashion syndrome, etc.

Modern pathology is not imaginable without the concept of syndrome, in the same way that the nomenclature of the syndrome seems more acceptable with the proper name of the author who first described it.

The number of syndromes inevitably increases from year to year in such a way that it is very difficult to have a complete view. There are so many that at the present time we can speak of a science, syndromatology (Schönfeld), or better we would say, syndromology (Llanio).

However, the concept or definition of syndrome has gone through different stages of which we will only point out some details.

Concept definition:

Hippocrates (460-377 BC) used the word syndrome to indicate a univocal group of regularly associated symptoms.

Galen (129-199 AD) used the term syndrome or symptom complex interchangeably.

Thomas Sydenham (1624-1689) considered the term syndrome similar to what years later was called a disease. In the following years these two meanings were delimited, although currently it is sometimes not easy in many cases.

In our opinion, it should be called a disease or nosological entity when it is exactly known that its etiology is unique and always presents with the same characteristics. However, there are different criteria in this regard and it is not our intention to spend time and space arguing or defining the exact concept.

The names that have been assigned to the syndromes are also dissimilar and sometimes capricious (as happens with diseases), so we can point out:

1. With the name of the author who described it (eponym): Libman-Sacks syndrome, Werner syndrome.
2. With the patient's name or initial: Harnutp syndrome, syndrome C, described by Opitz and collaborators in 1969, the first case in a family with the surname Camus.
3. With common terms from somewhere: kwashiorkor syndrome, which derives from kwashi = child and orkor = pink, a term used by the indigenous people of the Gold Coast because this condition is accompanied by malnutrition and pigment migration in such a way that black hair acquires a pink color.
4. According to etiology: withdrawal syndrome, arriboflavinosis syndrome.
5. According to the pathogenesis: Dumping syndrome, salt deficiency syndrome.
6. For the main symptom: anxiety or anxious syndrome.
7. By anatomical location: chiasmaic syndrome, middle lobe syndrome.
8. Due to the main anatomical lesion: cerebral sclerosis syndrome, aseptic epiphyseal necrosis syndrome.

9. By a word formed with the initials of the main symptoms: VATER syndrome, LEOPARD syndrome, HELLP syndrome, CRET syndrome, TAR syndrome.
10. By geographical location: Meleda syndrome (island in the Adriatic Sea where it was first observed), Tangier syndrome.
11. For religious reasons: Jobs syndrome. Which is based on a biblical precept that says: "Satan stigmatized Job with grains from the soles of his feet to the crown of his head."
12. Named after characters from novels: Munchaussen syndrome, people with psychiatric disorders who remember the famous character Baron Munchaussen, famous because he always lied and invented feats in which he had participated.

In this book we will maintain the authors in the designation of the syndromes in general without entering into the discussion between the defenders and detractors of the eponyms, although in others we will use the name best known to doctors.

It is good to remember that medicine is not the only science that uses eponyms. Biologists use them although with more systematization. Chemists to name new elements (parisium, californium). Physicists to name their laws or phenomena. Cartographers or explorers to name countries, towns, rivers, etc. (America, Alexandria). Astronomers to name the planets and stars.

However, confusion has been created in medicine and many are against eponyms due to the lack of systematic codification in this field.

Currently there are two currents in the terminology to designate pathological entities: scientific (still complex and confusing), which follows the scholastic method to try to categorize, give an entity its classification and finally a number; and the other, less scientific method, which designates an entity by the name of the discoverer, that of the patient or any of the variants listed above, or simply by a capricious decision.

It can be considered that the eponym has been useful in medical literature in all these years and has provided a double benefit: firstly, to honor and remember its discoverer as a respectful and well-deserved tribute to such distinguished authors, and secondly to facilitate its identification to remember the different syndromes and symptoms, which otherwise would require writing a more or less long sentence to name the syndrome, which has the consequence that sometimes there are so many symptoms indicated in its name that the syndrome is almost described.

In support of the defenders of eponyms, it can be added that they have had and continue to play an important role, as they allow the humanistic aspect of medicine to be perpetuated.

The learning that writing this book meant has been a great lesson for the authors and in particular for me, who has written or reviewed all the syndromes several times. We have enjoyed the anecdotes related to the syndromes and their origins, let us cite just two examples: the story of the Dutch admiral (Baron Wassenaer) who in 1879, during a copious meal in which he also drank a lot of alcohol, had a vomiting crisis and due to the efforts he made, his esophagus was seriously injured and he began to expel blood, after which he declared "I am going to die soon", which actually happened. His doctor Hermann Boerhaave described it so accurately the first time that it has been named after him ever since (Boerhaave syndrome). The case of Albatross syndrome, based on the fable of Samuel Taylor Coleridge (Rhyme of the Ancient Mariner) who tells the story of a sailor who killed one of those birds, which have never harmed anyone, and how he suffered for many years until he completely repented, which excited Johnstone and collaborators to give that name to certain cases of gastrectomy due to ulcer, performed by surgeons who have not taken into account a series of personal situations of the patients and carry for many years with the condemnation, demanded by these patients, whose sufferings have worsened.

For all the above about the syndrome, we consider that the doctor who has knowledge and masters this subject, symbolically has the key to be able to penetrate the mysterious, infinite and exciting world of medicine.

Prof. Raimundo Llanio Navarro

BRAIN ABSCESS-CARDIO-CONGENITAL PATHY SYNDROME

Synonymy

S. of paradoxical embolism.

Syndromography

Clinic

It can occur at any age, in patients with cardiac anomalies and right-to-left shunt, headache, stiff neck, lethargy, and fever.

Physical examination. Hemiplegia, aphasia, Jacksonian seizure, coma. Other signs of intracranial expansile injury or meningitis.

Paraclinical examinations

Blood. Polyglobulia, leukocytosis.

Cerebrospinal fluid. Increased tension, increased protein (if there is meningitis, increased leukocytes).

Skull radiology.

Angiography.

Echocardiogram.

Syndromogenesis and etiology

Acute infectious process of the central nervous system in patients with congenital heart anomaly and paradoxical embolism.

Literature

Ballet, Gilbert: "Des absces du cerveau consecutifs a certaines malformations cardiacaques." Arch. Gen. de Méd., 145:659-667, 1880.

Chambers, W.R.: "Brain abscess associated with pulmonary arteriovenous fistula." Ann. Surg., 141:276-277, 1955.

Karre, JR: Pathological Researches. Essay I. On malformations of the human heart; illustrated by numerous cases, and preceded by some observations on the method of improving the diagnostic part of medicine. London, Longman, 1814.

ASCENDING AORTA ANEURYSM SYNDROME PRESSING ON THE PULMONARY ARTERY

Synonymy

S. compression of the pulmonary artery by aneurysm of the ascending aorta.

Syndromography

Clinic

It occurs in syphilitic patients.

Physical examination. Aortic insufficiency, right ventricular hypertrophy, single second heart sound.

Paraclinical examinations

Radiology. Cardiomegaly. Great dilation of the pulmonary artery. Decreased pulmonary circulation, in one lung more than the other. Calcification of the aneurysm.

Electrocardiogram. Right axial deviation, vertical position, biventricular hypertrophy.

Cardiac catheterization. Left deviation of the right ventricle. Difficulty pushing the tip of the catheter into the compressed pulmonary artery.

Angiography. It demonstrates compression and the absence of valvular stenosis.

Blood. Positive serology.

Echocardiogram.

Syndromogenesis and etiology

It is due to a syphilitic aneurysm of the ascending aorta that compresses the pulmonary artery.

Literature

Kukarni, T.P.; M.J. Gandhi and K.K. Datey: "The syndrome of compression of the pulmonary artery by an aneurysm of the ascending aorta." Amer. Heart J., 65:678-682, 1963.

Pearson, J.R. and E.S. Nichol: "The syndrome of compression of the pulmonary artery by a syphilitic aortic aneurysm resulting in chronic cor pulmonale Qf, with a report of a case." Ann. Int. Med. 34:483-492, 1951.

AORTA ANEURYSM SYNDROME - VENA CAVA FISTULA

Synonymy

S. perforation of an aneurysm of the abdominal aorta into the inferior vena cava. S. of aorta-inferior vena cava fistula.

Syndromography

Clinic

This syndrome manifests itself during the fifth, sixth and seventh decades of life, predominantly in men, with abdominal, back and leg pain, shock, fever, and dyspnea.

Physical examination. Edema of the lower limbs and trunk, pulsating abdominal mass, systodiastolic or continuous abdominal murmur, slightly painful depressible abdomen, occasionally ascites.

Paraclinical examinations

Abdominal radiology. Calcification of the wall of the aneurysm.

Blood. Whiz retention.

Abdominal ultrasound.

Angiography.

Computerized axial tomography.

Syndromogenesis and etiology

The syndrome is caused by a perforated atherosclerotic aneurysm in the inferior cava.

Literature

Beall, A.C.. Jr.; D.A. Coorley: GC. Morris, et al.: Perforation of arteriosclerotic aneurysms into inferior vena cava." Arch. Surg., 86:809-818, 1963.

Syme, J.: "Case of spontaneous varicose aneurysm." Edimb. Med. Surg. J., 36:104-105, 1931.

AORTA DISsecting ANEURYSM SYNDROME

Syndromography

Clinic

It occurs before the age of 40. It is more common in men, between 40 and 70 years old. It is characterized by intense anterior chest pain that is relieved only with high doses of morphine.

Intense from the beginning and not progressive as in acute myocardial infarction. It radiates to the back, abdomen, lower limbs, neck and head. In some cases the pain may disappear. Shock, coma, hemiplegia, confusion, weakness in the legs.

Physical examination. Absence or inequality of carotid, brachial, and femoral pulses, which coincides with the pain or occurs a few hours after the pain. Pulsation in the sternoclavicular joint: Distension of the neck veins, systolic or diastolic murmur in 10-15% of cases. Elevation of blood pressure in contrast to signs of shock, pallor, tachypnea.

Paraclinical examinations

Electrocardiogram. Left ventricular hypertrophy, nonspecific changes, pericarditis.

Radiology. Elongation of the aorta.

Blood. Anemia, leukocytosis, hyperbilirubinemia.

Urine. Red blood cells, albumen, cylinders.

Syndromogenesis and etiology

The dissecting aneurysm of the aorta is caused by a half-cystic necrosis of the arterial wall.

In some cases it is associated with pregnancy, coarctation of the aorta, aortic stenosis, myxedema, atheromatous lesion of the aorta, aortic abscess.

Literature

Baer, S. and H. Goldburg: "The varied clinical syndromes by dissecting aneurysms." Amer. Heart J., 35:198-211, 1948.

De Bakey, M.E.; W.S. Henley: D.A. Cooley, et al.: "Surgical management of dissecting aneurysms of aorta." J. Thorac. Cardiov/. Surg., 49:130-149, 1965.

Hirst, A.E.; VJ. Hohns, Jr. and S.W. KIme, Jr.: "Dissecting aneurysm of aorta: a review of 505 cases." Medicine, 37:217-279, 1950.

UNSTABLE ANGINA SYNDROME

Synonymy

S. of coronary insufficiency. S. of acute coronary insufficiency. Prethrombotic S. S. intermediate coronary. S. of impending myocardial infarction.S. pre-infarction.

Syndromography

Clinic

This syndrome is more common in men (leaders, executives) with a history of diseases such as hypertension, diabetes, obesity or symptoms suggestive of cardiovascular disease.

Presenting symptoms: chest pain such as that of angina or heart attack or definite disease, generally substernal or parasternal or precordial, or abnormally localized and radiating to the arm, epigastrium, jaw or back.

Dyspnea, palpitations, fatigue, paresthesias, vomiting, flatulence.

Suspicious manifestations of the syndrome may be identified, such as: prolonged attacks at rest (successive or progressively increasing); intensification (with pattern changes) of pain in established angina; anginal pain at rest after a pain-free period in case of an old infarction.

Paraclinical examinations

Blood. Leukocytes, erythrocyte sedimentation rate and glutamicoxalacetic transaminase: normal or slightly

Chest radiology. Normal heart.

Electrocardiogram. Ischemic pattern almost always present (95%), S-T displaced well below, T abnormal.

Syndromogenesis and etiology

It is due to incomplete atherosclerotic obstruction and spasm of the coronary arteries.

It is a precursor syndrome to a myocardial infarction.

SYNDROME OF THE AORTA RIDING ON THE TABIOUE AND STENOSIS OF THE PULMONARY ARTERY EMERGING FROM THE LEFT VENTRICLE

Clinic

Early severe cyanosis. Little decreased physical resistance.

Physical examination. Grade I systolic tip murmur.

Reinforcement of the second lung sound. Growth of left cavities. Clubbing fingers on hands and feet.

Paraclinical examinations

Chest radiology. Front view: medium concave arch. Oblique view: disappearance of the concavity that is replaced by the prominence of a vessel whose anterior contour is tangent to the anterior edge of the underlying ventricle. Growth of left cavities.

Simultaneous catheterization and angiography.

Electrocardiogram.

Syndromogenesis and etiology

It is due to a congenital anomaly.

Literature

De la Cruz, M.V.; and J. P. Rocha: "An ontogenic theory for the explanation of congenital malformations involving the truncus and conus." Amer. Heart J., 51:782-805, 1956.

De la Cruz, M.V.; J. Espino-Vela and G. Anselmi: "Biventricular transposed aorta and left ventricular stenotic pulmonary artery, And embryologic and pathologic account a new syndrome whose existence was previously theoretically predicted." Amer. Heart J., 59:902-9 12, 1960.

CERVICAL AORTA SYNDROME

Syndromography

Clinic

Symptoms of vascular ring, diplegia, dyspnea, stridor, metallic cough, usually occurring in late childhood or adolescence.

Physical examination. A pulsating mass was palpated on the right side of the neck above the clavicle with murmur and thrill. When the mass is compressed, there is a marked decrease or disappearance of the femoral pulses.

Paraclinical examinations

Esophagram.

Angiography. Rarely necessary.

Echocardiography.

Syndromogenesis and etiology

It is caused by an anomaly of the aortic arch.

Literature

Beaven, T.E.D. and L. Fatti: Ligature of aortic arch in the neck". Brit. J. Surg., 34:414-416, 1946.

Massumi, R.; L. Wiener; L. P. Charif: "The syndrome of cervical aorta: report of a cases and review of the previous cases." Amer. J. Cardiol., 11:678-685, 1963.

AORTIC ARCH SYNDROME

Synonymy

S. of thoracocervical occlusive disease. S. of pulseless disease. Martorell's inverted coarctation. S. of chronic subclavian-carotid obstruction.

Syndromography

This syndrome includes:

1. S. of occlusion of the carotid artery.
2. S. occlusive of the vertebro-basilar artery.
3. S. occlusive of the subclavian artery.
4. S. of the subclavian theft.

Clinic

Although each syndrome shows a particular pattern, there is a variable degree of overlap and lesions of more than one artery, as well as congenital anatomical variations that can make differentiation confusing. In tables 1 and 2/Jones and collaborators try to differentiate symptoms and signs:

TABLE1. Symptoms associated with the involved artery

Symptoms	Vertebral Artery		Subclavian
	Unnamed		
Motor and sensory impairment	x		
Crisis	x		
Visual impairment	x		
Diplopia	x		
Aphasia-		x	
dysphasia	x	x	
Changes in memory			
Delirium	x	x	
Vertigo		x	
Arm			
None			x

TABLE2. Physical signs related to the artery involved

Signs	Artery		
	Unnamed	Subclavian	Vertebral
Blood pressure			
Reduction (in the arm) 160-30 mm Hg	x		x
Changes in pulse			
Arm	x		x
Neck			
Noises in the neck			x
Neurological deficit			
Ischemic changes in Subclavian			x
steal hand	x		x

Paraclinical examinations

Angiography. It is necessary to perform it on the aorta and the three indicated vessels in order to determine which one or which the lesion is located in.

Syndromogenesis and etiology

Atherosclerosis is responsible for the majority of cases with aortic arch syndrome. Nonspecific arteritis constitutes 16%; traumas, embolisms, constitute a group known as Takayasu syndrome; dissecting aneurysm, thoracic foramen syndrome, congenital stenosis (including that associated with supravalvular stenosis of the aorta), syphilis, pseudoxanthoma elasticum, polyarteritis nodosa, radiation.

Literature

Rotman, M.: “Aortic arch syndrome secondary to radiation in childhood. ” Intern Arch. Med., 124:87, 1969.

Thompson, B.W., et al.: "Aortic arch syndrome." Arch. Surg., 98:107, 1969.

TRAUMATIC ASPHYXIA SYNDROME

Synonymy

S. of egymotic mask. S. of traumatic cyanosis.

Syndromography

Clinic

This syndrome manifests itself at any age. It follows violent compression of the chest and/or abdomen.

Physical examination. Diffuse ecchymoses on the face, neck and upper chest. Vitreous, retinal, conjunctival hemorrhage (constant), associated with visual disorders, exophthalmos, mydriasis. Shock- that accords with the violence of the trauma.

Paraclinical examinations

Chest radiology.

Syndromogenesis and etiology

It is due to diapedesis of blood caused by a sudden and violent compression of the vascular system within the thorax or abdomen.
domen.

LOW EXPENDITURE SYNDROME

Syndromography

Clinic

Symptoms of generalized hypoperfusion: incoherent speech, obtundation, dyspnea, oliguria.

It is seen in heart failure, especially after heart surgery or myocardial infarction.

Paraclinical examinations

Hemodynamic tests. Cardiac output below 3 L/min/m². O2 consumption, lactic acid production, plasma catecholamines, blood gases.

Syndromogenesis and etiology

It is due to low cardiac output (below 3Lminm with evidence of peripheral vasoconstriction and tissue hypoperfusion caused by heart muscle disease.

Literature

Dietzman, R.H.; A. Ersek; C.W. Lillehei et al.: "Low output syndrome, recognition and treatment" J. Thoracic & Cardiovas. Surg.. 57:138-150, 1969.

Lillehei, R.C.; RH. Dietzman and J.H. Block: Hypotension and low output syndrome following cardiopulmonary bypass. In Norman, J.C., ed.: Cardiac Surgery. New York, Appleton- -Century-Crofts, 1967, pp. 437-456.

BERNHEIM SYNDROME

Synonymy

S. of obstructive insufficiency of the right ventricle. S. of obstruction and insufficiency of the right ventricle.

Syndromography

Clinic

In this syndrome there are two stages: 1, few or no symptoms: 2a, dyspnea may be absent: 2b, total heart failure, dyspnea, orthopnea, etc.

Physical examination. Two stages: 1, right atrium enlargement, jugular venous engorgement, increased venous pressure, normal circulation time; 2a, venous engorgement systemic, clear lungs, hepatomegaly, edema, possible ascites, normal or slightly increased circulation time. 2b, total heart failure.

Paraclinical examinations

Electrocardiogram.
Circulation time.
Chest radiology.
Fluoroscopy.

Syndromogenesis and etiology

The mechanism that causes symptoms is left ventricular hypertrophy; The growth and protrusion of the ventricular septum into the right ventricle obstructs blood flow to the right atrium.

The most frequent causes of this syndrome are: aortic stenosis and high blood pressure.

Literature

Bernheim, P.: “De l'Astolie Veineuse dans l'hypertrophie du coeur gauche par stenose concomitante du ventricule droit". Rev. de Med. Paris, 30: 785-800, 1910.

Drago, E.E. and J.T. Aquilina: “Bernheim's syndrome". Amer, J. Cardiol., 14:568-572, 1964.

POST-CARDIOTOMY BIPHASIC SYNDROME

Syndromography

Clínica

Its onset is after cardiac surgery; A few days after surgery, the first phase appears, characterized by fever and mouth lesions (small painful, non-ulcerated indurations) that disappear in 2 weeks. The second phase occurs after 3-6 weeks and is characterized by hepatosplenomegaly, pericardial and pleural friction rub. The course is benign due to its long duration (months). This syndrome appears to represent a combination of postperfusion syndrome and postpericardiotomy syndrome.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Kahn, D.R.; P.Y. Ertel, W.H. Murphy, et al.: "Pathogenesis of the postcardiotomy syndrome." J. Thorac. Cardiov. Surg., 54:682-687, 1968.

BLAND-WHITE-GARLAND SYNDROME

Synonymy

S. of the left coronary artery that arises from the pulmonary artery.

Syndromography

Clinic

According to some authors, this syndrome has 4 phases:

Phase I. Neonatal, normal appearance of the child at birth and until shortly after.

Phase II. Transient, death or survival according to the development of adequate intercoronary anostomosis, distress, symptoms and signs of myocardial ischemia.

Phase III. Progressive improvement of symptoms and signs of ischemia. Frequently left chamber dilation and secondary mitral insufficiency, with continuous murmur. Later, arrhythmias.

Phase IV. Coronary artery steal syndrome, in this phase sudden death is common in adolescents or adults.

Other authors describe 4 types that are generally similar to the phases described: infantile syndrome, mitral insufficiency, continuous murmur syndrome, sudden death in adolescents or adults.

Paraclinical examinations

Electrocardiogram. Inverted T wave in DI and DII, deep Q wave in DI; signs of anterolateral infarction

Chest radiology. Cardiomegaly at the expense of the left ventricle, sometimes pulmonary edema.

Retrograde aortography. Makes the diagnosis.

Cardiac catheterization.

Syndromogenesis and etiology

It is due to a congenital malformation in which the emergence of the left coronary artery in the pulmonary artery is confirmed.

Literature

Abrikossoff, A.: "Aneurysma des linken Uerzventrikels mit abnormer abgangstelle der linken Koronararterie von der Pulmonalis beieinen funfononatlichen Kinde." Virchow's Arch.
Path. Anat. Berl.. 203:413-420, 1911.

Bland, E.E: PS. White and J. Garland: "Congenital anomalies of coronary arteries. Report of unusual case with cardiac hypertrophy." Am. Heart J., 8:787, 1933.

Friedberg, C.K.: Diseases of Heart, Philadelphia, Saunders, 1966.

Gasul, M.G.: R.A. Arcilla and M. Lev: Heart diseases in children.
Ed. Pitman, Med. London, 1966. p.976.

Gland, E.E, et al.: Congenital anomalies of the coronary arteries, report of an unusual case associated with cardiac hypertrophy".
Amer. Heart J., 8:787-801, 1932-1933.

BOUVERET-HOFFMAN SYNDROME

Synonymy

S. of idiopathic paroxysmal atrial tachycardia. S. of paroxysmal atrial tachycardia.

Syndromography

Clinic

This syndrome occurs at any age, most frequently between the second and fourth decades of life.

In childhood it affects boys almost exclusively.

The onset is sudden, without warning, occasionally precipitated by an emotion or a sudden change in position.

Suddenly there is a sensation of a blow in the chest, apparent cardiac arrest, precordial discomfort, palpitations.

Occasionally: 1, feeling of pain in the throat, pulsation in the neck; 2, anxiety, weakness, coldness or sweating may accompany the crisis; 3, gastrointestinal symptoms, epigastric discomfort, abdominal distension, heartburn, nausea, vomiting; 4, polyuria during or at the end of the crisis.

Physical examination. Tachycardia. Only in very prolonged crises or in the presence of an underlying illness, can signs of congestive heart failure appear. Small and fast pulse. Blood pressure may fall (especially systolic with concomitant decrease in pulse pressure).

Paraclinical examinations

Electrocardiogram. Fast and regular ventricular complexes with T waves often difficult to identify and slightly modified by fusion with P waves.

Syndromogenesis and etiology

The etiology is obscure; occasionally this syndrome is associated with biliary diseases, or with patients with Wolf-Parkinson-White syndrome. There are no known congenital forms.

Literature

Bouveret, L.: "De la tachycardie essentielle paroxystic." Rev. of Med., 9: 753-793, 837-855, 1889.

Friedberg, C.K.: Diseases of Heart. Philadelphia, SaunderS, 1966.

HUNTER CANAL SYNDROME

Syndromography

Clinic

This syndrome is characterized by the following symptoms in the lower limbs: 1, intermittent claudication: 2, disappearance of the pulse; 3, decrease or disappearance of arterial oscillations.

Syndromogenesis and etiology

The syndrome is caused by circulatory insufficiency caused by periarterial fibroadhesive lesions at the level of Hunter's canal.

ALCOHOLIC CARDIOMEGALY SYNDROME WITH EMPHYSEMA

Syndromography

Clinic

It is observed exclusively in chronic alcoholic men.

Patients present with chronic bronchitis, slowly progressive cardiopulmonary failure.

Paraclinical examinations

Blood. Anemia.

Determination of pyruvic acid and function tests hepatic.

Respiratory functional tests.

Electrocardiogram.

Chest radiology.

Syndromogenesis and etiology

The most frequent causes are: alcohol ingestion, secondary lung infections as a complicating factor.

cation.

Literature

Josserand, A.: “Le syndrome gros coeur et emphyseme important complications viscerale de l'alcoholisme et facteur de surmortalite masculine.” Lyon Medical, 189:42-44, 1953.

IDIOPATHIC CARDIOMYOPATHY SYNDROME

Synonymy

S. of idiopathic myocardial hypertrophy. of non-coronary cardiomyopathy. S. of adult fibroelastosis. Subendocardial fibroelastosis syndrome (Becker syndrome). S. of chronic pernicious myocarditis. S. of cardiovascular collagenosis. Nutritional heart disease. S. de Abramov-Fiedler.

Syndromography

Clínic

It predominates in men in 78% of cases, with an average age of onset of 34 years. The manifestations are:
easy fatigue, dyspnea on exertion, palpitations, malleolar edema (especially in the afternoons), progressively orthopnea and paroxysmal nocturnal dyspnea, syncope on exertion and pain in the right upper quadrant.

Physical examination. Cardiomegaly, normal first sound, accentuation of the second lung sound (when other signs of congestion develop), pathological third sound, occasionally murmur of secondary mitral or tricuspid regurgitation, hepatomegaly, hepatojugular reflux.

Paraclinical examinations

Electrocardiogram. Various abnormal patterns.
Chest radiology. Cardiomegaly, pulmonary congestion.
Echocardiogram.

Syndromogenesis and etiology

Multiple etiologies have been considered. Many cases are a consequence of infectious myocarditis.

Bibliography

Fiedler, A.: Uber akute interstiielle Myokarditis. In Festsc hrift zur Feier des funfzigjahrigen Bestehens des Stadtkranken hauses zu Dresden-Friedrichstadt. Dresden: W. Baensch, 1899, pp. 1-20.

Sanders, V.: "Idiopathic disease of myocardium." Arch. Int. Med., 112; 661--676, 1963.

Wenger, H.N.: Rare Cause of Heart Diseases." In Hurst, J.W., and R.B. Logue, ed.: The Heart. New York, McGraw-Hill, 1966, p. 916.

AMYLODOSIC HEART DISEASE SYNDROME

Syndromography

Clinic

It occurs in both sexes. It generally begins between the fourth and fifth decades of life (in familial cases). There is dyspnea on exertion and fatigue.

Physical examination. Progressive signs of right heart failure. Normal or slightly decreased blood pressure. Hepatomegaly frequently disproportionate to the degree of heart failure.

Splenomegaly (occasional).

Paraclinical examinations

Blood. Normal, there are no changes in proteins or polysaccharides.
Electrocardiogram.
Congo red test.
Rectal biopsy.
Chest radiology.
Bone marrow. Increase in plasma cells.

Syndromogenesis and etiology

The syndrome is caused by amyloid infiltration into the myocardium.

It is transmitted by autosomal dominant inheritance (described in a family in Denmark). In other cases it adopts indefinite forms of inheritance or occurs sporadically.

The etiology is genetic.

Literature

Frederiksen, T.; H. Gotzsche; N. Harboe, et al.: "Familial primary amyloidosis with severe amyloid heart disease." Amer. J. Med., 33:328-348, 1962.

Layman, T. E.; B.W. Lyne and J.E. Edwards: “Systemic amyloidosis with cardiac involvement suggesting coronary arterial disease." Geriatrics, 23:103-112, 1968.

RADIATION HEART DISEASE SYNDROME

Syndromography

It can take different clinical forms that are indistinguishable from those produced by other causes:

1. Acute pericarditis (during radiation or after some time).
2. Chronic pericarditis (with effusion, or constriction, associated with endomyocardial fibrosis).
3. Coronary heart disease with myocardial infarction.
4. Mitral regurgitation and cardiomyopathy.

Syndromogenesis and etiology

Damage to the heart and pericardium occurs from high doses of radiation in chest cancer.

Literature

Stewart, J.R.; K.E. Cohn; L..F. Fajardo, et al.: “Radiation induced heart disease, a study of twenty-five patients.” Radiology, 89:302-310, 1967.

SHOCK SYNDROME

Synonymy

S. of circulatory collapse. of shock.

Syndromography

Clinic

There are manifestations of nausea, weakness, coldness, restlessness, confusion and sometimes loss of consciousness.

Physical examination. Generalized skin pallor, with cyanotic areas. Decrease in temperature with cold limbs, moist skin, profuse sweating, weak, depressible and tachycardic pulse. There is arterial hypotension below 70 for the maximum and 40 for the minimum, which is also manifested by clinical signs: absence of the venous network and superficial arterioles. Muffled heart sounds, sometimes imperceptible. In the final phase: acute myocardial failure and/or ventricular fibrillation.

Three phases have been described in the shock according to its intensity:

1. Latent or compensated shock phase.
2. Confirmed or decompensated shock phase.
3. Irreversible shock phase.

Paraclinical examinations

Blood. Blood hemoconcentration and leukocytosis, hyperglycemia and decreased serum cholinesterase activity. When the condition progresses: hypoglycemia, acidosis, hyperkalemia, tissue

Syndromogenesis and etiology

The syndrome can be produced by different mechanisms:

1. Hypovolemic shock: hemorrhages, burns, dehydration, heat stroke, etc.
2. Septic shock: diffusion of bacteria and/or toxins, mainly gram-negative germs (septicemia, intestinal or uterine perforation, puerperal sepsis).
3. Cardiogenic shock: inflow heart failure as seen in myocardial infarction.
4. Shock due to inadequate cardiac filling: severe tachycardia, pericardial tamponade, tension pneumothorax. In which there are two variants: decreased diastolic filling (tachycardias) and mechanical obstruction (those mentioned above).
5. Shock due to mechanical obstruction of central circulation: pulmonary thromboembolism, with its intense reflex reactions.
6. Other shock: anaphylactic, acute adrenal insufficiency, etc.

Literature

LLanio, R. and colab.: Clinical Propedeutics and Physiopathology. 3rd ed. Editorial Pueblo y Educación, City of Havana, 1996.

SCIMITAR SYNDROME

Synonymy

S. of anomalous venous drainage of the right lung into the inferior vena cava.

Syndromography

Clínica

It predominates in females, and the diagnosis is made more frequently in the third decade of life. It can be completely asymptomatic or slightly symptomatic. It is characterized by fatigue, dyspnea, history of pneumonia, cough, chest pain, wheezing. Rarely growth retardation.

Physical examination. Heart displaced to the right, rarely decreased size and mobility of the right hemithorax, rales, loud heart murmurs over the sternum or on one side of it, occasionally the physical examination is negative.

Paraclinical examinations

Chest radiology. Heart displaced to the right;
venous trunk abnormally located, parallel to the right border of the heart, widened and curved in its middle and anterior part (scimitar sign); opacity of the right lung.

Fluoroscopy. Absence of pulsation.

Tomography.

Bronchoscopy.

Bronchography. Right bronchiectasis.

catheterization.

Respiratory functional tests.

Angiography

Echocardiography.

Syndromogenesis and etiology

It is due to a congenital malformation. There are familial cases (autosomal dominant inheritance).

Literature

Chassinat, R.: "Observations d'anomalies anatomiques remarquables de 1' appareil circulatoite avec hépatocele congenitale, symptom particulier." Arch. Ge. Med., 11:80-91, 1836.

Cooper, G.: "Case of malformation of the thoracic viscera: consisting of imperfect development of right lung and transposition of the heart." London Med. Gaz.. 18:600-602, 1836.

Kiely, B.; J. Filler; S. Stone, et al.: "Syndrome of anomalous venous drainage of the right lung to the inferior vena cava; a review of 67 reported cases and three new cases in children."
Amer. J. Card., 20: 102-116, 1967.

McKusick, V.A. and R.N. Cooley: "Drainage of the right pulmonary veins into the interior vena cava." New English J. Med., 252:291, 1955.

Neill, C.A.; C. Ferencz; D.C. Sabiston et al.: "The familial occurrence of hypoplastic right lung with systemic arterial supply and venous drainage: ' scimitar syndrome'". Bull. Johns Hopkins.
Hosp. 107, 1960.

Winslow (1739), cit. by Stoermer, J. in Opitz-Schmid.: Pediatric Encyclopedia (Spanish trans.) t. VII. Ed. Morata, Madrid, 1968.

INTERMITTENT CLAUDICATION SYNDROME

Syndromography

Clínica

It is manifested by dullness, weakness and painful stiffness of one or both lower limbs, after some time of walking, it disappears with rest.

Syndromogenesis and etiology

Vascular syndrome caused by partial arterial obstruction, generally due to arteriosclerosis.

EISENMENGER COMPLEX SYNDROME

Synonymy

S. de Eisenmenger.

Syndromography

Clínica

Dyspnea on exertion, delayed physical development, repeated lung infection with occasional hemoptysis, which implies a poor prognosis. Cough can be the dominant symptom in older
late

Physical examination. In childhood: moderate cardiomegaly, thrill at the left edge of the sternum, systolic murmur, diastolic dragging. In adolescence and early adulthood:
cyanosis, clubbing of fingers on hands and feet.

Paraclinical examinations

Electrocardiogram. Deviation of the electrical axis to the left in young children and to the right in older children.

Shortened circulation time.

Chest radiology. Right ventricular enlargement, prominent pulmonary artery trunk as well as the central branches with amputation of the peripheral branches.

Cardiac catheterization. Increased pressure in the pulmonary artery, increased oxygen pressure in the right ventricle.

In older children, decreased oxygen saturation index.

Echocardiogram.

Blood. Polyglobulia.

Angiocardiography. Inverted or bidirectional shunt.

Syndromogenesis and etiology

It is due to a congenital defect with a left-to-right shunt due to a ventricular and/or atrial septal defect or ductus arteriosus and/or atrial septal defect. When pulmonary hypertension is established, the shunt reverses from right to left.

Literature

Eisenmenger, V.: "Die angeborenen Defekte der Kommerscheidewand des Herzens." Ztschr. F. Klin. Med., suppl. 32:1-28, 1897.

Rees, R.S.O., et al.: "The Eisenmenger syndrome." Clin. Radiol., 18:366, 1967.

Wood, P: The Eisenmenger syndrome". Brit. Med. J. 2:701-709, 1958.

POST-PEREUSION PULMONARY CONGESTION SYNDROME

Synonymy

S. of pulmonary postperfusion vasculitis.

Syndromography

Clinic

It occurs in patients operated on for cardiac diseases when cardiopulmonary bypass techniques are used, and is characterized by fever and dyspnea that occur immediately after the surgical intervention.

Physical examination. Cyanosis, hypotension, signs of pulmonary edema.

Paraclinical examinations

Chest radiology. Diffuse opacity of both lung fields.

Syndromogenesis and etiology

The mechanisms that cause this syndrome are:

1. Fall in mean arterial pressure.
2. Anoxia.
3. Reaction to the destruction of blood elements, resulting from turbulence in the pumping system.
4. Reaction to remaining protein substances in the heart-lung machine from previous perfusions.

Literature

Kahn, D.R.; P.H. Ertel; W.H. Murphy, et al.: Pathogenesis of the postcardiotomy syndrome." J. Thorac. Cardiov. Surg., 54:682-687, 1967.

Kreel, I.; L.I. Zaroff; J.W. Canter, et al.: "A syndrome following total body perfusion." Surg. Gynec. Obstet., 111:317-321, 1961.

Reyman, TA.: “Postperfusion syndrome". Amer. Heart J., 72:116-123, 1966.

ACUTE CORPULMONALE SYNDROME

Synonymy

S. of pulmonary thromboembolism.

Syndromography

Clinic

This extremely serious syndrome begins suddenly with sudden or medium intensity dyspnea, cough (sometimes with hemoptoic sputum), precordial tightness without syncope, paleness, and symptoms of circulatory collapse (arterial hypotension, tachycardia, weak pulse, peripheral vasoconstriction).

Pain of medium or great intensity that simulates a heart attack (not constant), which is replaced by intense chest pain (especially during inspiration), of a pleuritic type, when a thrombus reaches the cortical portion of the lung. Sometimes there is hemoptoic sputum. Sometimes sudden death.

Physical examination. In addition to those already mentioned, there may be clinical signs of pulmonary condensation associated or not with signs of liquid interposition. Sometimes it is accompanied by acute right heart failure with right gallop and/or acute lung edema. Arterial hypotension, engorgement of the neck veins, cyanosis and shock.

Mortality is high and occurs in the first 30 minutes to 1 hour after the onset of symptoms. Sometimes there is sudden death.

Sometimes the symptoms are not so alarming, they depend on the massiveness of the embolism.

Paraclinical examinations

Electrocardiogram. SI, QII, right axial deviation, incomplete right bundle branch block, pulmonary P, sometimes atrial fibrillation.

Central venous pressure. Increased.

Radiological study. Sometimes diaphragmatic elevation, increased caliber of vessels, peripheral clear areas after vascular occlusion (Westermarik sign), sometimes pleural effusion.

Pulmonary arteriography.

Perfusion scintigraphy.

Syndromogenesis and etiology

This syndrome is due to an acute decompensation of the right ventricle caused by an increase in pressure in the pulmonary artery, which is produced by massive pulmonary embolism of any etiology. There are various degrees according to vascular obliteration, with a decrease in minute volume. The most common is acute fulminant symptoms with sudden death.

The risk factors for phlebothrombosis are: immobilization, surgical interventions, trauma, obesity, oral contraceptives (especially associated with smoking), pregnancy, treatment with corticosteroids or diuretics, malignant tumors, protein C deficiency.

Etiology. It is caused by embolisms that almost always come from the lower limbs, but also from tumors, fat embolisms due to fractures, etc.

GOUT HEART SYNDROME

Synonymy

S. with a suspended heart. S. Evans and Lloyd-Thomas.

Syndromography

Clinic

It occurs in all ages and both sexes, with chest pain and/or palpitations.

Physical examination. Generally, patients have an asthenic habit and do not present evidence of cardiovascular disease.

Paraclinical examinations

Electrocardiogram. ST depression in DIII (especially on inspiration), also in DII even less, without change when the patient stands up.

Radiology. In frontal view, normal location of the heart appears, but in deep inspiration the left cardiophrenic angle moves towards the center; In right anterior oblique view the heart and diaphragm are widely separated; In the left anterior oblique, a space is observed between the heart and the diaphragm.

Syndromogenesis and etiology

It is caused by an alteration in the position of the heart that appears elongated and vertical (pendant or dripping heart). It is common in patients with a certain constitutional type (Stiller's asthenic habit).

Literature

Evans, W. and H. G. Lloyd-Thomas: The syndrome of the suspended heart", Brit. Heart. J., 19:153-158, 1957.

HYPERKINETIC HEART SYNDROME

Synonymy

S. of increased ejection. S. with the heart of an athlete. (See mitral valve prolapse syndrome.)

Syndromography

Clínica

It is observed in all ages. Less than 50% of patients have cardiorespiratory symptoms (dyspnea, orthopnea, chest discomfort), about 20% participate in athletic activities.

Physical examination. Hyperactivity of the heart and great vessels, full and rapid pulse, systolic ejection murmur at the base or left edge of the sternum, intense. 50% of patients present a systolic ejection click and splitting of the second lung sound. Femoral venous murmur that increases when raising the legs or with exercise. Blood pressure lability (50%).

Paraclinical examinations

Electrocardiogram. Left ventricular hypertrophy. In some patients completely normal.

Chest radiology. Generally normal. Pulmonary plethora only in a minority of cases.

Syndromogenesis and etiology

Possible psychological tension, anxiety, prehypertension state. Some similarity is observed with the stimulation effect of catecholamines. Ejection time shorter than normal. Stroke volume not necessarily increased. It is currently considered in mitral valve prolapse syndrome.

The etiology is unknown.

Literature

Gorlin, R.: "The hyperkinetics heart syndrome." J.A.M.A., 182:823-829, 1962.

Starr, I. and L. Jonas: “Supernormal circulation in resting subjects (hyperkinemia) with a study of the relation of the kinemic abnormalities to the relation of the kinemic abnormalities to the basal metabolic rate.” Arch. Int. Med., 71:1-22, 1943.

Strano, A.; L. Di Renzi and V. Pennelti: "On the perioheral venous murmur of the circulatory hyperkinetic syndrome'". Angiology, 17:213-222, 1966.

RIGID HEART SYNDROME

Synonymy

S. of hemodynamic restriction. S. of restrictive heart disease.

Syndromography

Clinic

Dyspnea on exertion that can reach orthopnea, occasionally chest pain.

Physical examination. Systemic venous hypertension, pulsus paradoxus, Kussmaul's sign may be present, dull or normal heart sounds, third diastolic sound (of great importance in the diagnosis), hepatomegaly, edema, ascites.

Paraclinical examinations

Electrocardiogram. Normal or with some alteration.

Chest radiology. Contributes to differential diagnosis.

Cardiac catheterization.

Echocardiogram.

Other exams. According to the suspected etiology.

Syndromogenesis and etiology

Chronic constrictive pericarditis of any cause.

Primary and secondary cardiomyopathies. Amyloidosis.

Hemochromatosis.

It is currently considered that one third of heart failure is due to diastolic dysfunction (stiff heart) and the main mechanism is a delay in relaxation or decreased filling of the left ventricle.

The factors that cause decreased filling of the left ventricle due to altered muscle compliance are:

1. High blood pressure.
2. Acute myocardial ischemia.
3. Diabetes mellitus.
4. Obesity.
5. Hypertrophic cardiomyopathy.

Literature

Kilpatrick, T.R.; H.M. Horack andC. B. Moore: «Stiff heart' syndrome; an uncommon cause of heart failure". Med. Clin. N. Amer., 51:959-966, 1967.
Shabetai, R.: N.O. Fowler and J.C. Fent: Restrictive cardiac dis- ease, pericarditis and the myocardiopathies." Amer. Heart J., 69:271-280, 1965.

COSTOCLAVICULAR SYNDROME

Synonymy

Falconer-Weddell S. S. of military posture.

Syndromography

Clinic

Pain, paresthesia, weakness in arms and hands.

Physical examination. Edema, venous congestion in the arms, weak pulses.

Syndromogenesis and etiology

This syndrome occurs by holding the shoulders back and down as in the military posture, when the patient is carrying heavy packages on their back.

Literature

Falconer, M.A. and G. Weddell: Costoclavicular compression of subclavian artery and vein: relation to scalenus anticus syndrome. Lancet, 2:539-543, 1943.
Spring, M.: "Cervical syndrome." Hosp. Me., 2:18-28, 1966.

IDIOPATHIC PULMONARY ARTERY DILATION SYNDROME

Syndromography

Clinic

The patients are asymptomatic.

Physical examination. The findings on auscultation and in the phonocardiogram are characteristic: splitting of the first sound at the tip with the first component of equal or greater intensity than the second. Systolic click that gives way to a loud systolic ejection murmur at the pulmonary focus.

Widely split second noise. A short diastolic murmur may be present in the second intercostal space.

Paraclinical examinations

Electrocardiogram. Normal.
Chest radiology. Dilation of the pulmonary artery.
Phonocardiogram.
Echocardiogram.
Cardiac catheterization. Normal right ventricular systolic pressure with a difference across the valve of 15 mm Hg or less during exercise, which has diagnostic value.

Syndromogenesis and etiology

It could be an incomplete form of Marfan syndrome. The diagnostic criteria for this rare form would be: 1, simple dilation of the pulmonary artery or its main branches; 2, occasional association with aortic hypoplasia; 3, absence of congenital heart disease, lung disease, rheumatism and syphilis.

Literature

Ramsey, H.W.; A. De la Torre; J.W. Linhart, et al.: "Idiopathic dilation of the pulmonary artery." Amer. J. Cardiol., 20:324-330, 1967.
Wessler, H., and L. Jackes: Clinical Roentgenology of Disease of the Chest Troy (N.Y.), Southworth, 1923, pp. 26-29.

PAPILLARY MUSCLE DYSFUNCTION SYNDROME

Synonymy

S. of the papillary muscle.

Syndromography

Clinic

It generally occurs in patients over 40 years of age, without specific symptoms. Possible symptoms of the disease responsible for the dysfunction of the papillary muscle: angina, congestive heart failure.

Physical examination. Apical “diamond” systolic murmur that radiates to the axilla, sometimes with thrill, which decreases in intensity if there is dilation of the left ventricle and congestive insufficiency. The murmur is fixed although the lesions evolve.
Absence of end-systolic click.

Paraclinical examinations

Electrocardiogram. Type I: moderate depression of the J point with ST concave upward or slightly convex downward.
Type II: slight to moderate depression of the J point, with very convex upward ST and inversion of T. Type III: marked depression of the J point, with slight upward convexity of the beginning of ST.

Phonocardiogram.
Echocardiogram. A flapping mitral valve could be observed.

Syndromogenesis and etiology

This syndrome can be caused by different causes:

1. Circulatory failure: angina, infarction, systemic circulatory disorders.
2. Dilation of the left ventricle: localized, generalized.
3. Non-ischemic atrophy of the papillary muscle.
4. Defective development, endocardial disease.
5. Heart muscle disease.
6. Functional disorders of the papillary muscle, rupture.

Literature

Burch, G.E.; N.P. DePasquale and J.H. Phillips: "Clinical manifestations of papillary muscle dysfunction." Arch. Int. Med., 112:112-117, 1963.
Burch, GE.; N.P. DePasquale and J.H. Phillips: The syndrome of papillary muscle dysfunction." Amer. Heart J., 75:399-415, 1968.

SINUS NODE DYSFUNCTION SYNDROME

Synonymy

S. of the diseased sinus node. S. of the sick breast.

Syndromography

Clinic

Patients present symptoms of periodic cerebral arterial insufficiency: personality changes, irritability, memory loss, sleep disturbance, digestive disorders, transient oliguria and in severe cases paresis and impaired judgment.

Physical examination. Sudden, persistent, severe sinus bradycardia. Cessation of sinus rhythm for short intervals with the appearance of other rhythms; long periods of sinus arrest resulting in cardiac arrest, episodes of slow ventricular rhythm in chronic atrial fibrillation. Difficulty recovering sinus rhythm after cardioversion.

Paraclinical examinations

Electrocardiogram.

Syndromogenesis and etiology

Symptoms are due to cerebral, renal and cardiac hypoperfusion.

This syndrome is caused by a temporary injury to the breast caused by catheterization. Destruction of the sinoatrial node tissue due to ischemia, sclerosis or inflammation.

It can be evidenced by the use of drugs:
digitalis, propranolol, morphine sulfate and lidocaine.

Literature

Ferrer, M.I.: The sick sinus syndrome in atrial disease".
J.A.M.A., 206:645-646, 1968.

ABNORMAL PULMONARY VENOUS DRAINAGE SYNDROME WITH MITRAL STENOSIS

Synonymy

Partial anomalous pulmonary venous drainage with stenosis mitral.

Syndromography

Clinic

Similar to that of atrial septal defect with tight mitral stenosis.

Paraclinical examinations

Electrocardiogram. Right ventricular hypertrophy.

Chest radiology. Enlarged right ventricle, hypoplastic aorta.

Cardiac catheterization. High wedge pulmonary pressure, large shunt from left to right, high oxygen saturation in the right atrium. The finding of high or low pulmonary wedge pressure in different parts of the lungs could be additional evidence of the presence of partial anomalous venous drainage.

Syndromogenesis and etiology

It is due to a congenital defect.

Literature

Hughes, C.W. and P.C. Rumor: Anomalous pulmnonary veins".
Arch. Path., 37:364-366, 1944.

Wassermil, M. and M.S. Hoffman: "Partial anomalous pulmonary vein drainage associated with mitral stenosis with an intact atrial septum." Amer. J. Cardiol., 10:894-899, 1962.

DRESSLER SYNDROME

Synonymy

S. of post-myocardial infarction.

Syndromography

Clinic

It occurs 2-10 weeks after a recent heart attack in 3% to 4% of cases. Fever, chest pain.

Physical examination. Pericarditis, pleurisy and pneumonitis with a tendency to recurrence. However, the complete picture (including pericarditis and pneumonia) is not always present.

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation rate, leukocytosis. Autoantibodies against the heart muscle.

Blood culture.

Chest radiology.

Electrocardiogram and echocardiogram. The results are as expected in any spill.

Syndromogenesis and etiology

Evidence has been found that an antigen produced by myocardial necrosis induces the formation of autoantibodies and these would be responsible for pleuropulmonary lesions.

The etiology is unknown.

Literature

Dressler, W.: "A complication of myocardial infarction resembling idiopathic recurrent benign pericarditis." Cyclulation, 12:697, 1955.
Dressler, W.: "A post-myocardial infarction syndrome." J.A.M.A., 160:379, 1956.

Levin, E.J., et al.: "Dressler syndrome (postmyocardial infarction syndrome)." Radiology, 87:731, 1966.
Markoff, R.: “Klnische Bedeutung des Dressler-Syndroms.”
Deustach. Med. Wschr., 93:627-633, 1968.

EBSTEIN SYNDROME

Synonymy

S. of Ebstein's tricuspid valve anomaly. S. of Ebstein's malformation.

Syndromography

Clinic

Asymptomatic for years or dyspnea and fatigue.

Physical examination. Cyanosis at birth or later, or recurrent with increasing severity. Clubbing (moderate) fingers on hands and feet. Arrhythmia crisis.

Marked cardiomegaly. The three or four beat rhythm is characteristic. Systolic murmur at the left edge of the sternum, at the tip. In some cases also a diastolic murmur at the tip.

Paraclinical examinations

Electrocardiogram. Right bundle branch block, and prolonged PR, high P waves, atrial tachycardia, premature beats and frequent flutter.

Fluoroscopy and radiology. Enlargement of the right atrium, small pulmonary artery, decreased pulmonary circulation.

Angiography. Very enlarged right atrium.

Cardiac catheterization. Increased pressure in the right atrium, evidence of shunts from right to left and vice versa.

Eeocardiogram.

Syndromogenesis and etiology

It is due to a congenital malformation with low implantation of the tricuspid in the right ventricle and deformity of the leaflets.

Literature

Ebstein, W.: Ueber einen sehr seltenen Fall von Insufficenz der Valvula tricuspidalis, bedingt durch eine angeborene hochgradige Missibildung derselben". Arch. Anat. Physiol., 238:254, 1866.

Hippo, F.A. and S. Arthachinta: “Ebstein's anomaly of the tricuspid valve, a report of 16 cases and review of literature". Program Cardiov. Dis., 7:434-448, 1965.

POSTACHYCARDIA ELECTROCARDIOGRAPHIC SYNDROME

Syndromography

Clínic

It appears after an attack of paroxysmal tachycardia (for example, Wolff-Parkinson-White syndrome). The electrocardiographic alterations are reversible and can last between 6 and 60

Paraclinical examinations

Electrocardiogram. Inverted T in one or more depressed ST leads where the former is more important. QTprolonged.

Syndromogenesis and etiology

The etiology is unknown.

MIMA POLYMORPHA ENDOCARDITIS SYNDROME

Syndromography

Clínic

This syndrome occurs in young men with no history of heart disease. The onset is acute with chest pain and slight fever.

Physical examination. Tachycardia, diastolic aortic murmur, maculopapular rash, occasionally embolic phenomenon, splenomegaly in one out of five patients and arthralgias in two out of five.

Paraclinical examinations

Blood. Usually normal blood count. Erythrosedimentation moderately elevated or normal. Negative blood culture or reports an atypical gram-negative organism (Mima polymorpha).

Syndromogenesis and etiology

Endocarditis due to Mima polymorpha or Herellea vaginicola (gram-negative pleomorphic germs) belonging to the Mimeae group.

Literature

Shea, D.W. and J.H. Phillips: "Mimeae endocarditis; a clinical syndrome"' Amer. J. Med. Sci., 252:201-205, 1960.

STRAIGHT BACK SYNDROME

Synonymy

S. flat chest.

Syndromography

Clinic

False heart disease due to deficiencies in the diagnosis, which occurs in the activities and work life of the patients. In some cases there is dyspnea.

Physical examination. Loss of dorsal kyphosis (flat palm of the examiner's hand on the spine does not cause normal deviation of the fingers).

Cardiac auscultation: systolic ejection murmur, late, harsh, at the base, grades I-IV. Reinforced second lung sound.

Paraclinical examinations

Frontal and lateral chest radiology. Absence of the anterior concavity of the upper dorsal region of the spine, with compression of the heart against the sternum and displacement of great vessels.

Echocardiogram. Normal.

Electrocardiogram. Normal.

Syndromogenesis and etiology

Congenital anomaly of the upper dorsal column that reduces the anteroposterior diameter of the thorax and causes false cardiomegaly and murmurs. In some cases, pulmonary obstruction with pulmonary hypertension.

This syndrome is seen more frequently in patients with congenital heart disease, especially atrial septal defects.

Literature

Leinbach, R.C.; J.W. Harthorne and R.E. Dinsmore: "Straight back syndrome with pulmonary venous obstruction." Amer. J. Cardiol., 21:588-592, 1968.

Ravlings, M.S.: The 'straight back' syndrome. A new cause of pseudo-heart disease." Amer. J. Cardiol., 5:333-338, 1960.

Twigg, H.H.; B.C. of Leon; JK. Perloff, et al.: "The straight back syndrome; radiographic manifestations". Radiology, 88:274-277, 1967.

LOWER LIMB STASIS SYNDROME

Syndromography

Clínica

It is characterized by: pigmentation (due to hemosiderin deposits), eczema, skin ulcerations, edema (de-

insufficient reabsorption of interstitial fluid by the lymphatics), lymphedema, cellulitis, indurated thrombophlebitis.

Syndromogenesis and etiology

Complication secondary to the presence of varicose veins chronicles.

COMPLICATED SUPRAVALVULAR STENOSIS SYNDROME

Synonymy

S. of supravalvular aortic stenosis. S. of supravalvular aortic stenosis and childhood hypercalcemia. S. de Beuren.

Syndromography

Clinic

This syndrome affects both sexes. It begins at birth or at an early age. It is characterized by eating disorders, anorexia, vomiting, slow weight gain, and delayed physical and mental development. Variable degrees of hypotonia, easy fatigue, chest pain and occasional syncope. Decreased exercise tolerance.

Physical examination. Weight and height below the third percentile. Facial abnormalities: elfin face (broad forehead, thick cheeks, pointed chin). Hypertelorism, flattened nasal bridge. Bilateral corneal opacity; hypoplasia of tooth enamel with malocclusion and cavities. Blood pressure inequality between both arms.

Heart: harsh systolic ejection murmur, with more intense thrill in the second intercostal space. Absence of aortic systolic click, occasionally systolic aortic murmur.

Paraclinical examinations

Blood. Hypercalcemia (common) or normal calcium and phosphorus. Hypercholesterolemia (occasional). Hypercalcemia and hypercholesterolemia usually disappear in the second or third decades of life.

Radiology. Metaphyseal osteosclerosis and craniowestenosis.

Angiocardiography. It shows an apparent diaphragm in the trunk of the pulmonary artery, reminiscent of a donut seen from the side, in the area between the valve and the supervalvular diaphragm.

Cardiac catheterization.

Electrocardiogram.

Urine. Decreased endogenous creatinine clearance.

Chromosome study. Normal pattern.

Syndromogenesis and etiology

Primary intrauterine disorder. Family tendency (no evidence of genetic basis). Possible disorder of vitamin D metabolism.

The etiology is unknown.

Literature

Kostis, JB., et al.: The syndrome of supravalvular aortic stenosis: Report of two unusual cases with comments on the etiology". Chest, 57:253, 1970.

Martin, E.C., et al.: "Supravalvular aortic stenosis." Br. Heart J., 35:758, 1973.

Roberts, N., and C.A.F. Moes: "Supravalvular pulmonary stenosis." J. Pediatr., 82:838, 1973.

Williams, J.C.P.: B.G. Barratt-Bayes, and J.B. Lowe: "Supravalvular aortic stenosis." Circulation, 24:1311-1318, 1961.

UNCOMPLICATED SUPRAVALVULAR STENOSIS SYNDROME

Synonymy

S. of supravalvular stenosis of the aorta.

Syndromography

Clinic

It is much more common in men; history of cyanosis in childhood.

Physical examination. Inequality between the right and left pulses in the upper limbs and neck; high systolic murmur at the base, not systolic click or diastolic murmur. Normal facies and intelligence. Congestive heart failure may develop.

Paraclinical examinations

Electrocardiogram.

Cardiac catheterization.

Chest radiology.

Echocardiogram.

Selective angiography of the right ventricle.

Syndromogenesis and etiology

Congenital malformation: 1, sporadic, non-familial?

2, incomplete form of infantile hypercalcemia syndrome?

The etiology is unknown.

Literature

Antia, A.U.; H.E. Wiltse; R.D. Rowe, et al.: Pathogenesis of the supravalvular aortic stenosis syndrome." J. Pediat., 71:431-441, 1967.

Archer, R.S.: *Note on a congenital band streching across the origin of the aorta". Dublin J. Med. Sci., 65:405-406, 1878.

Logan, W.E; E.W. Jones; E. Walker, et al.: "Familial supravalvular aortic stenosis." Brit. Heart J., 27:547-559, 1956.

ENDOCARDINAL FIBROELASTOSIS SYNDROME

Synonymy

S. de Weinberg-Himelfarb. Subendocardial sclerosis S.

S. of endocardiodysplasia.

Syndromography

Clinic

This syndrome occurs in children between 6 weeks and 6 months old, occasionally in older children. It is manifested by dyspnea, vomiting, and edema of the lower limbs.

Physical examination. Tachycardia, weak heart sounds, gallop rhythm, rales, hepatomegaly. Cyanosis crisis. Poor development with little weight gain.

Paraclinical examinations

Electrocardiogram. Left ventricular hypertrophy, sometimes complete left bundle branch block.

Angiocardiography.

Chest radiology and fluoroscopy. Pulsatile cardiac edge, weak and undulating contraction.

Echocardiogram.

Syndromogenesis and etiology

Inherited congenital defect of the elastic tissue of the heart or damage from hypoxia in some circumstances. It is transmitted by autosomal recessive inheritance; once dominant with reduced penetrance.

Literature

Kreysing, FL.: Die Krankheiten des Herzens systematische bear-beiltet. Ed. Maurer, Berlin, 1815.

Moller, S.H.; R.0. Fisch; A.H.L. From et al.: "Endocardial fibroclastosis occurring in a mother and son." Pediatrics, 38:918, 1966.

Pérez Díaz, L.: Aortic coarctation in infants and children and its relationship with endocardial fibroelastosis. Thesis of the Univ. Autonomous Community of Madrid, 1974.

Süll, WJ.S.: Endocardial fibroelastosis. Amer. Heart J. 61:579-585, 1961.

Weinberg, T. and A.J. Himelfarb: "Endocardial Fibroelastosis (so-called fetal endocarditis)". Bull. J. Hopk. Hosp., 72:299- -306, 1943.

SPONTANEOUS AORTOCAVA FISTULA SYNDROME

See S. of traumatic aortocaval fistula

Syndromography

Clinic

It predominates in men (no cases have been reported in women), in ages between 40 and 78 years. Pain in the back and abdomen occasionally radiating to the lower limbs.

Dyspnea, anorexia, nausea, vomiting, restlessness, confusion, lethargy, stupor, oliguria, shock.

Physical examination. Palpable aneurysm, systolic or only systolic abdominal murmur, cardiomegaly, edema in lower limbs, venous pulse, ascites, hepatomegaly.

Wide differential pressure due to lowering of the minimum.

Marked increase in venous pressure in lower limbs

rivers.

Paraclinical examinations

Urine. Hematuria.

Stool. Blood.

Blood. Hyperazoemia.

Syndromogenesis and etiology

The syndrome is caused by the rupture of an aortic aneurysm in the vena cava.

The causes may be arteriosclerosis, syphilis, Marfan syndrome, mycosis.

Literature

Nenhaus, H.P. and H. Javid: "The distinct syndrome of spontaneous abdominal aorto-caval fistula." Amer. J. Med., 44:464-473, 1968.

Syme, J.S. (1831) quoted by Boffi, L.: Rottura di aneurisma dell'aorta addominale nella vena cava inferiore (rassegna della letteratura e study clinico). Chir. e pat. sper., 3:73-94, 1955.

TRAUMATIC AORTOCAVA FISTULA SYNDROME

Syndromography

Clinic

Pain in the back or abdomen occasionally radiating to the lower limbs. Dyspnea, oliguria, anorexia, nausea, vomiting.

Physical examination. Palpable aneurysm, systodiastolic murmur, pulmonary rales, cardiomegaly, edema, hepatomegaly, ascites.

Paraclinical examinations

Chest radiology.

Aortography.

Syndromogenesis and etiology

The cause is trauma.

Unlike spontaneous aortocaval fistula, the prognosis is better and patients can survive even without surgical treatment for an average of 5 years.

Literature

Nennhaus, H.P. and H. Javid: "The distinct syndrome of Spontaneous abdominal aortocaval fistula." Amer. J. Med.. 44:464-473, 1968.

PHLEBECTASIA SYNDROME OF THE JEJUNUM, MOUTH AND SCROTAL

Syndromography

Clinic

Symptoms of peptic ulcer and occasionally symptoms of gastrointestinal bleeding.

Physical examination. Caviar-like spots on the tongue, Fordyce angiokeratoma on the scrotum, mane.

Paraclinical examinations

Radiology. Stomach, duodenum, intestinal transit.

Stool. Searching for hidden blood.

Gastrochymogram.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Miller, D.A. and W.A. Akers: "Multiple phlebectasia of the jejunum, oral cavity, and scrotum." Arch. Int. Med., 121:180-182, 1968.

Rappaport, I. and M. A. Schiffman: "Multiple phlebectasia involving jejunum, oral cavity, and scrotum." J.A.M.A., 185:437-440, 1963.

PHLEBODYNIA SYNDROME

Synonymy

S. of venous pain.

Syndromography

Clinic

It predominates in women and appears epidemically, as severe pain along the superficial and deep veins of the legs. Moderate general symptoms: malaise, headache and light fever.

Physical examination. Pain in the affected limb without redness or heat in the venous tract, sensation of swelling (diseased wall or thrombus). Positive Homan sign, moderate increase in volume of one or both legs.

Paraclinical examinations

All negative.

Syndromogenesis and etiology

The possibility of mass hysteria is a phenomenon to consider.

The etiology is unknown.

Literature

Brosius, G.R.; M.D. Calvert and T.D.Y. Chin: “Epidemic phlebodynia.” Arch. Int. Med., 108:442-447, 1961.
Pearson, J.S.: “Phlebodynia a new epidemic (?) disease.” Circulation, 7:370-372, 1953.

ALBA DOLENS PHLEGMASY SYNDROME

Synonymy

S. of painful puerperal edema. Femoral thrombophlebitis.

Syndromography

Clinic

It occurs in the postpartum period or as a post-surgical complication and is characterized by pain, paleness and increased volume of the legs; general malaise.

Physical examination. Edema and coldness in the affected limb.

Syndromogenesis and etiology

It is due to thrombosis of the uterine veins that extends to the veins of the lower limbs.

Literature

Allen, E.V.; N.W. Barker and E.A. Hines: Peripheral Vascular Diseases. ed. 3, Philadelphia, Saunders, 1962.

CERULEA DOLENS PHLEGMASY SYNDROME

Synonymy

S. of gangrene of venous origin. S. of massive acute venous obstruction.

Syndromography

Clinic

Usually sudden pain in all toes, occasionally gradual following recurring episodes.

always severe venous thrombosis. In some cases it is the progressive evolution of phlegmasia alba dolens.

Physical examination. A clear line of demarcation is observed between the affected area and healthy tissue, increased volume and cyanosis, color in discolored areas. Peripheral pulses are palpable, strong, and full in early stages until obliterated by progressive edema. In some cases the manifestations cover the entire sole of the foot in the form of a sheath.

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation, increase in urea nitrogen.

Syndromogenesis and etiology

The syndrome is idiopathic or can frequently be associated with malignant diseases. It is due to an interruption of blood flow through the capillary bed determined by venous thrombosis and edema.

Bibliography

Hueter, C.: Fall von Grangran in Folge von Venebliteration", (Virchows) Arch. path. Anat., 17:482-488, 1859.
Loewenthal, J. and J. May: "Phlegmasia cerulea dolens." Brit. J. Surg., 52:584-587, 1965.

GALLAVARDIN SYNDROME

Synonymy

S. of blochypnea. S. of inhibited breathing.

Syndromography

Clinic

It is triggered by exercise or varying degrees of emotional tension. There is an inhibition of breathing, rest produces relief. It is not accompanied by panting or dyspnea. In some cases it alternates with typical symptoms of angina.

Physical examination. Negative

Paraclinical examinations

Laboratory tests. All negative.

Electrocardiogram. It may show a pattern of angina especially after exertion.

Syndromogenesis and etiology

It is an equivalent of the anginal syndrome in patients with coronary atherosclerosis, alteration of the central nervous system (spinothalamic bundle to the respiratory center in stimulation of the spinal cord).

The cause appears to be an increased susceptibility to stimulation of the respiratory center.

Literature

Gallavardin, L.: "Y a-t-il un equivalent non douloureux l'angine de poitrine d' effort?". Lyon Med., 134:345-358, 1924.
Roesler, H.: “Blockypnea (inhibited respiration), an equivalent of the angina syndrome." Amer. J. Med. Sci., 244:85-87, 1962.

HEBERDEN SYNDROME

Synonymy

S. of angina pectoris. S. of stenocardia. S. de Elsner.
S. of Rougnon de Magny.

Syndromography

Clinic

This syndrome begins suddenly and can be moderate, severe or distressing. It is characterized by dull, oppressive precordial pain, accompanied by anxiety. Irradiated to the left shoulder, arm and hand. Short in duration (generally less than 3 min), precipitated by emotion, effort, exposure to cold, heavy eating. It is relieved with nitroglycerin (see also Gallavardin syndrome).

Physical examination. Negative.

Paraclinical examinations

Electrocardiogram. In crises there is depression of the ST segment, outside of them, this is generally normal.
During the effort alterations may appear.

Radiology. Negative, in advanced disease there may be cardiomegaly.

Syndromogenesis and etiology

It is caused by myocardial hypoxia as a consequence of decreased coronary flow due to atherosclerosis. Very rarely it is due to another anatomical or functional cause.

Literature

Anonymnous: The Ebers Papyrus, XXVII, p. 47. Translated by Ebbell, B., Copenhagen, Levin and Munksgaard, 1937.
Friederg, C.H.: Diseases of the Heart. ed. 3 th, Philadelphia, Saunders, 1966.
Heberden, W.: "Some account of a disorder of the breast." Med. Tr. Roy. Coll. Phys., London, 2:59-67, 1772.

RIGHT HEART HYPOPLASIA SYNDROME

This name is used to designate a group of congenital cardiac anomalies that have atresia or anatomical stenosis of the right hemicardium, with hypoplasia of the right ventricle and hypertrophy of the left and functionally a shunt from right to left, generally at the atrial level. This group includes: tricuspid atresia, pulmonary atresia, severe pulmonary stenosis with intact ventricular septum and congenital isolated hypoplasia of the right ventricle (rare).

Literature

Morgan, A.D., T.G. McLoughlin; T.D. Bartley, et al.: "Endocardial fibroelastosis of the right ventricle in the newborn. Presenting the clinical picture of the hypoplastic right heart syndrome." Amer. J. Cardiol., 18:933-937, 1966.

IDIOPATHIC ORTHOSTATIC HYPOTENSION SYNDROME

Synonymy

S. of orthostatic hypotension. S. of postural hypotension. (See S. of Shy-Drager.)

Syndromography

Clínica

It is more common in men (4:1), slow progression of symptoms, sometimes years before the patient seeks medical attention. Impotence is sometimes the first symptom.

It is manifested by episodic symptoms of exhaustion, lightheadedness, blurred vision, mental slowness and lassitude in an upright position. Syncope is common, preceded by the premonitory symptoms already described. Seizures may occur (especially if trying to get the patient up after a fall). There may be chronic diarrhea, nocturnal polyuria, and heat intolerance.

Physical examination. Patients generally have a youthful appearance. The pallor precedes the attack. Decrease in systolic and diastolic pressures in the upright position. The Valsalva maneuver causes a marked drop in blood pressure and the characteristic bradycardia (present in normal individuals) is absent. A variable degree of anhidrosis may be revealed by sweat tests.

Paraclinical examinations

Special tests. Vasopressin increases systolic and diastolic pressures in patients (not in normal individuals).

Sweat tests.

Basal metabolism. Low.

Blood. Moderate increase in urea nitrogen, slight anemia.

Syndromogenesis and etiology

A probable mechanism is the dysfunction of the autonomic nervous system that would cause: defective arteriolar constriction, anhidrosis, xerostomia, heat intolerance, impotence.

The etiology is unknown.

Literature

Bradbury, S. and C. Eggleston: Postural hypotension: A report of three cases." Amer. Heart J., 1:73-86, 1925.
Lanbry, C. and E. Doumer: “L'hypotension orthostatique.” Press. Med., 1:17-20, 1932.
Levin, J.M.; P. Ravenna and M. Weiss: "Tdiopathic orthostatic hypotension." Arch. Int. Med., 114:145-148, 1964.

HOLT ORAM SYNDROME

Synonymy

S. of familial defect of the heart and upper limbs. S. of atriodigital dysplasia. S. cardiobrachial.

Syndromography

Clinic

The complete syndrome presents: congenital defect of the heart and upper limbs. Isolated upper limb heart defect can be seen in individuals from the same family. It may be asymptomatic or present cardiac symptoms such as dyspnea, fatigue.

Physical examination. Typical of heart disease: atrial septal defect, rarely interventricular (ostium secundum), and anomalies of the upper limbs that include polydactyly and syndactyly. The association of the defect of the members

Upper brows of the "radial type" (digitized thumb simulating another index finger, absence of the thumb on one side, with hyperphalanx of the thumb on the other, hand vara, short radius, and radioulnar synostosis) with heart defect is much more common than that of the "ulnar type." Occasionally: absence of pectoral muscle, funnel chest, scoliosis, cleft lip and palate, genital anomaly.

Paraclinical examinations

Special tests. Evaluation of the heart defect.

Skeletal radiology.

Chromosome study. Normal, although abnormalities of chromosome 16 have been reported.

The study of dermatoglyphs only shows abnormalities secondary to the musculoskeletal defect and they do not appear genetically determined.

Electrocardiogram. QR elongation.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance with variable expressivity.

The etiology is genetic.

Literature

Holt, M. and S. Oram: Familial heart disease with skeletal malformations." Brit. Heart J., 22:236-242, 1960.

Kaufman, R.L.; D.L. Mc. Rimoin; W.H. Alister and colbs: "Variable expression of the Holt-Oram syndrome". Am. J. Dis. Child., 127:21, 1974.

Massumi, R.A. and D.O. Nutter: The syndrome of familial defects of heart and upper extremities (Holt-Oram syndrome)".
Circulation, 34:65-76, 1966.

Sánchez Cascos, A.: "A Holt-Oram syndrome". Paediat Act.
Scand., 56:313, 1967.

Zetteerovist, P.: The syndrome of familial atrial septal defect, heart arrhythmia and hand malformation (Holt-Oram) in mother and son", Acta Paedriat., 52:115, 1963.

HUGHES-STOVIN SYNDROME

Synonymy

S. of pulmonary artery aneurysms-pulmonary artery thrombosis-peripheral venous thrombosis.

Syndromography

Clinic

Headache, fever, cough, hemoptysis, papilledema.

Paraclinical examinations

Chest radiology. Dense shadow at the site of the aneurysm, decreased vascularization in different parts of the lung.

Angiocardiography. Aneurysms of the segmental branches of the pulmonary arteries.

Syndromogenesis and etiology

Thrombosis in leg veins, vena cava, jugular vein or right heart associated with pulmonary artery aneurysms.

Literature

Hughes, J.P. and P.G. Stovin: "Segmental pulmonary aneurysm with peripheral venous thrombosis." Br. J. Dis. Chest, 53:19, 1959.

Wolpert, S.M.; PC. Kahn and K. Farbman: The radiology of the Hughes-Stovin syndrome." Am. J. Roentgenol., 112:383, 1971.

CIRCULATORY INSUFFICIENCY SYNDROME WITH INCREASED OUTPUT

Syndromography

Clínic

Symptoms predominantly of heart failure right.

Physical examination. Increased heart volume, increased venous pressure, generally decreased diastolic pressure, increased systolic and pulse pressure. All signs of congestive heart failure are present.

Paraclinical examinations

Special tests. Circulation time, decreased or normal. Increased venous pressure.

Syndromogenesis and etiology

There is a decrease in circulatory resistance caused by different conditions: arteriovenous fistula, hyperthyroidism, anemia, beriberi, severe pulmonary emphysema with hypoxemia, pregnancy, advanced Paget's disease and Albright syndrome. These alterations lead to heart failure.

Literature

Friedberg, C.K.: Disease of the Heart. ed. 3th, Philadelphia, Saunders, 1966.

PULMONARY VALVE INSUFFICIENCY SYNDROME

Syndromography

Clinic

This syndrome is characterized by the absence or debatable presence of clinical manifestations. There may be mild dyspnea on exertion, hyperdynamic contraction of the right ventricle.

Physical examination. Low-frequency diastolic murmur, with greater intensity at the pulmonary focus or a little lower along the left edge of the sternum, which increases in intensity during inspiration. A short, systolic ejection murmur may be present.

Paraclinical examinations

Electrocardiogram. Normal with ventricular hypertrophy right.

Chest radiology.

Echocardiogram.

Phonocardiogram.

catheterization.

Syndromogenesis and etiology

It is due to a congenital defect of the valve or idiopathic dilation of the pulmonary artery.

Literature

Kezdi, P.; W.s. Priest and J.M. Smith: Pulmonic regurgitation.
Quart. Bull. Northwest. Univ. Med. School, 29:368-373, 1955.

Criscitiello, M.G. and W.P. Harvey: "Clinical recognition of congenital pulmonary valve insufficiency." Amer. J. Card., 20:765-772, 1967.

POST RADIATION VASCULAR INSUFFICIENCY SYNDROME

Syndromography

Clínic

It appears late as a consequence of radiotherapy, especially of the pituitary in the case of acromegaly.

Physical examination. Symptoms of vascular obstruction with hemiparesis and other signs of decreased or interrupted circulation in the affected area.

Paraclinical examinations

Arteriography.

Syndromogenesis and etiology

Post-radiation vascular changes. (See also post-radiation brain necrosis syndrome.)

Literature

Darmondy, W.R.: L.M. Thomas and E.S. Gurdjian: Postirradiation vascular insufficiency syndrome." Neurology, 17:1190-1192, 1967.

JERVELL-LANGE-NIELSEN SYNDROME

Synonymy

S. cardioauditory. S. deafcardiac. S. long QT and deafness.

Syndromography

Clinic

It affects children of both sexes with bilateral deafness from infancy and childhood. Repeated attacks of syncope occur.

Attacks may begin with a high-pitched scream followed by paleness and later cyanosis and short-lived loss of consciousness followed by deep sleep. Attacks can result in sudden death due to ventricular arrhythmias and can be triggered by exercise or emotional states.

It is characterized by deaf-mute, dyspnea, nutritional disorders, growth retardation.

Physical examination. Hypertelorism, wide-based nose, low set of ears. Variable skeletal malformations: acrocephaly, scoliosis, pectus excavatum, syndactyly, polydactyly. Heart: signs of pulmonary stenosis and/or mitral regurgitation.

Paraclinical examinations

Electrocardiogram. QT prolonged. Ventricular arrhythmias may be found during attacks.

Audiometry. Bilateral loss of auditory perception.

Blood. Moderate hypochromic anemia.

Other examinations (electrocardiographic, electroencephalographic and biochemical). They do not show changes in this

Chest and skeleton radiology.

Chromosome study. Negative.

Cardiac catheterization.

Syndromogenesis and etiology

The mechanism of QT prolongation, as well as that of arrhythmias, is unknown. The delay in ventricular repolarization increases the vulnerable period and, therefore, the tendency to ventricular arrhythmias. The mechanical refractory period seems to predispose to arrhythmias.

It is transmitted by autosomal recessive inheritance. Consanguinity is common.

The etiology is genetic.

Literature

Forney, W.R; S.J. Robinson and D.J. Pascoe: "Congenital heart dyscase, deafness and skeletal malformations, a new syndrome?" J. Pediat., 68:14-26, 1966.

Jervell, A. and E. Lange-Nielsen: Congenital deaf mutism, functional heart disease with prolongation of Q-T interval and sudden death." Amer. Heart J., 54:59-68, 1957.

Lisker, S.A. and D. Finkelstein: "The cardioauditory syndrome of Jervell and Lange-Nielsen: report of an additional case with radioelectrocardiographic monitoring during exercise." Amer. J. Med. Sci., 252:458-464, 1966.

Meissner, E: Taubstummheit and Taubstummenbildung". Leipzig und Heidelberg, Wenter, 120, 1856.

KLIPPEL-TRENAUNAY-WEBER SYNDROME

Synonymy

S. of angiosteohypertrophy. S. of hypertrophic verrucous nevus. S. of hypertrophic hemangiectasia. St. Parkes-Weber

Syndromography

Clínic

Nevus located unilaterally on a limb. Varicose veins. Hypertrophy of the soft tissue and bones of the affected limb that makes it more elongated and warmer than the healthy one.

Varicose veins and osteohypertrophy are not always present at birth, but develop during the first months or years of life. It is frequently associated with polydactylism and syndactylism.

Paraclinical examinations

Radiology. Thickening of the cortex of the bones is observed, but this does not constitute an important diagnostic element.

Syndromogenesis and etiology

This syndrome is possibly caused by a hereditary weakness of the mesenchymal tissue of the walls of the

glasses.

It occurs sporadically, so its etiology has not been well defined.

Literature

Brooksaler, F.: The angio-osteohypertrophy syndrome." Amer. J. Dis. Child., 112:161-164, 1966.

Klippel, M. and P. Trenaunay: "Naevus variqueux osteohypertrop hique." Arch. Gen. Med. (Paris), 3:641-642, 1900.

Weber, P.E.: "Angioformation in connection with hypertrophy of limbs and hemihypertrophy." Brit. J. Derm., 19:23 1-235, 1907.

LERICHE SYNDROME

Synonymy

S. of thrombosis of the abdominal aorta. S. of gradual thrombosis of the terminal portion of the aorta. S. of occlusion of the aortic bifurcation.

Syndromography

Clinic

This syndrome is characterized by intermittent claudication of the lower limbs: pain and discomfort in the lower region of the body (muscles, hips or buttocks), weakness in the legs, paleness (in the standing position) in the feet and legs.

Physical examination. Arterial pulses in the legs decreased. two or absent; Occasionally, noises can be heard over the arteries: abdominal aorta, iliac and femoral arteries.

Muscle atrophy, skin generally normal or pale, cold, cyanotic, with trophic changes and gangrene in very advanced stages.

Paraclinical examinations

Oscillonmetry. Absence of pulses.

Abdominal radiology. Saddle calcification at the level of the aortic bifurcation.

Doppler. Decreased circulation.

Angiography. It allows determining the site and extent of the obstruction.

Syndromogenesis and etiology

It is caused by atheromatous plaques in the bifurcation of the aorta, segmental arteritis, gradual thrombosis of the terminal portion of the aorta.

Literature

Beckwith, R. et al. : *Chronic aortoiliac thrombosis: A review of sixty-five cases". New Engl. J. Med., 258:761, 1958.

Hurst, J.W.: Disease of Aorta. In Beeson, P.M., and McDermott, W., Cecil-Loeb Textbook of Medicine. ed. 12th, Philadelphia, Saunders, 1967.

Leriche, R.: "De la résection du carrefour aortico-iliaque avec double sympathectomie lombaire pour thrombose arterique de l'aorte. Le syndrome de lóblitération terminoaortique par arterite", Presse Med., 48:33-604, 1940.

LIBMAN-SACKS SYNDROME

Synonymy

S. of atypical verrucous endocarditis. S. of non-rheumatic verrucous endocarditis. Osler-Libman-Sacks S. S. by Kaposi-Besnier-Libman-Sacks.

Syndromography

Clínica

The symptoms are the same as those of SLE (systemic lupus erythematosus) (see).

Physical test: Apical systolic murmur.

Paraclinical examinations

Laboratory tests. Those indicated to confirm the LES.

Syndromogenesis and etiology

Endocarditis is due to the course of systemic lupus erythematosus.

Literature

Libman, E. and B. Sacks: “A hitherto undescribed form of valvular and mural endocarditis.” Arch. Int. Med., 33:701-737, 1924.

LUTEMBACHER SYNDROME

Syndromography

Clinic

It predominates in young women, with a history of rheumatic fever, or heart disease. The development of symptoms is slow.

Physical examination. Typical of mitral stenosis and intra-atrial communication. Atrial fibrillation in advanced ages.

Paraclinical examinations

Electrocardiogram. Binaural enlargement, right axis, right ventricular growth.

Chest radiology. Small aorta, dilated pulmonary artery, enlarged pulmonary cone, right hypertrophy.

Echocardiogram. Narrow mitral, communication interatrial

Angiography.

Cardiac catheterization.

Syndromogenesis and etiology

The syndrome is caused by a congenital atrial septal defect with added rheumatic mitral stenosis.

Literature

Lutembacher, R.: “De la stenose mitrale avec interauriculaire communication.” Arch. du Mal du coeur., 9:237-250, 1916.

Muller, W.H.; JB. Littlefield and J.R. Beckwith: "Surgical treatment of Lutembacher's syndrome." J. Thorac. Cardiov. Surg., 51:66-770, 1966.

PNEUMATIC HAMMER SYNDROME

Synonymy

Vibration S.

Syndromography

Clinic

It occurs in individuals exposed to handling vibrating tools. Exposure to cold produces a typical Raynaud's phenomenon (see this one). First it affects only the hand that is closest to the end of the implement, then all the fingers.

When the attack is severe, the sensitivity of the fingers can be completely lost.

Physical examination. During attacks, color change of the fingers, which is normal in the inter-seizure period. No trophic changes.

Paraclinical examinations

Laboratory tests. Those necessary to rule out other causes of Raynaud's phenomenon.

Syndromogenesis and etiology

Possible damage to the digital vessels with spasm on exposure to cold.

The etiology is unknown

Literature

Allen, E.V.; N.W., Barker and E.A. Hines: Peripheral Vascular Diseases. ed. 3, Philadelphia, Saunders, 1962.
Cottingham, C.C. quoted in the Presidents Monthly Report, The Stonecutter's J., 32:5, 1917.

MEADOW SYNDROME

Synonymy

S. of postpartum heart disease. S. of postpartum cardiomyopathy. S. of idiopathic myocardial failure in the puerperium.

Syndromography

Clinic

Symptoms appear between the second week and the second month after childbirth. It is characterized by cough, paroxysmal nocturnal dyspnea, hemoptysis, and chest pain.

Gastrointestinal symptoms: nausea, vomiting. Symptoms of cerebral and pulmonary embolism more frequently in multiparous black women.

Physical examination. The physical findings are those of congestive heart failure, first left and then right. Diastolic hypertension, alternating pulse, gallop rhythm, cyanosis, hepatomegaly. Normal eye fundus.

Paraclinical examinations

Chest radiology. Increase in the transverse diameter of the heart.

Electrocardiogram. Inversion of the T wave, pathological Q wave and conduction disorders.

Echocardiogram.

Syndromogenesis and etiology

The syndrome combines the clinical, hemodynamic and pathological characteristics of congestive cardiomyopathy.

It shows a tendency to relapse in subsequent deliveries.

Mortality is high.

It is considered due to myocarditis or cardiomyopathy of possible viral, autoimmune, or metabolic origin. Its family occurrence has been reported.

The etiology is unknown.

Literature

Friedberg, C.K.: Diseases of the heart. ed. 3, Philadelphia, Saunders, 1966, p. 1002.
Hull, E. and E. Hakkesbring: “Post-partal heart disease.” New Orleans Med. Surg. J., 39:550-557, 1937.
Meadow, W.R.: Tdiopathic myocardial failure in the last trimester of pregnancy and the puerperium." Circulation, 15:903-914, 1957.

MONDOR SYNDROME

Synonymy

S. of sclerosing periphlebitis of the chest wall.

Syndromography

Clínic

Predominates in women. It appears from the third to sixth decades of life. Little or no discomfort.

Physical examination. A reddish cord usually appears on the side of the chest that goes down the costal margin to the abdominal wall, adhered to the skin, but not deeply.

Syndromogenesis and etiology

Obliterans phlebitis of the thoracoepigastric vein. Frequent history of trauma.

Literature

Hogan, GF.: *Mondor's disease". Arch. Int. Med., 113:881-885, 1964.

Mondor, H.: "Tronculitis percutaneous subaigué of the pario thoraci que antérolatérale." Academic Mem. Chir. (Paris), 65:1271- -1278, 1939.

NONNE-MILROY-MEIGE SYNDROME

Synonymy

S. of hereditary trophoedema. S. of familial lymphedema. Milroy disease. Meige's disease. S. of hereditary tropholymphedema. S. of congenital elephantiasis. S. hereditary lymphedema I (Nonne-Milroy type). S. hereditary lymphedema II (type) Meige

Syndromography

Clinic

It predominates in females, from birth or early childhood. Its onset is gradual and asymptomatic.

Physical examination. Unilateral or bilateral malleolar edema that ascends to the knee and eventually higher; at the beginning of easy godet, it disappears with elevation of the leg. The skin is soft, firm and of normal color. Subsequently, the edema is hard, it does not disappear with elevation of the limb, the skin becomes rough, pigmented, at the site of the edema. As associated findings:
Congenital chylous ascites, intestinal lymphangiectasia with proteinorrhea, pleural effusion.

Syndromogenesis and etiology

Congenital autosomal dominant familial disorder. Lymphatic obstruction or contractility defect, absence of valves, weakness of the wall with extravasation.

The etiology is genetic.

Literature

Meige, H.: "Le trophoedeme chronique hereditaire." Nouv. Iconogr. (Salpetriere), 12:453, 1889.

Milroy, W.E: Underscribed variety of hereditary oedema." New York Med. J., 56:505-508, 1892.
Nonne, M.: «Vier Falle von Elephantiasis congenita herrkditaria». Arch. Path. Anat. (Paris) 125: 189-196, 1891.
Shirger, A.; E.G. Jr. Harrison and J.M. Janes: "Idiopathic lymphedema, review of 131I cases." J.A.M.A., 182:14-22, 1962.

PRECORDIAL PRESSURE SYNDROME

Syndromography

Clinic

It is observed in young healthy individuals, with no sex preference. Suddenly, a strong, imposing pain appears over the tip of the heart, almost always at rest and associated with inclined postures of the body, lasting about 2-3 minutes. Breathing is shallow to minimize pain, and position changes can ease pain. A deep breath can make it disappear.

It does not irradiate, recurrence is sporadic.

Physical examination. Negative. Patients with weak constitution median.

Paraclinical examinations

Laboratory tests. All negative.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Miller. A.J. and T.A. Texidor: "Precordial catch. a neglected syndrome of precordial pain." J.A.M.A., 159:1364-1365, 1955.

ORTNER SYNDROME

Synonymy

S. cardiovocal.

Syndromography

Clinic

Hoarseness occurs in the course of cardiovascular conditions: aortic arch injury, mitral stenosis, congenital heart disease, coronary heart disease.

Physical examination. Poor movement or paralysis of the left vocal cord.

Paraclinical examinations

Those indicated in the cardiovascular conditions that are present.

Syndromogenesis and etiology

It is due to an injury to the left laryngeal nerve, which is compressed between the aorta and the dilated pulmonary artery.

Literature

Ortner, N.: "Recurrenslabmung bei Mitraisienose", Wien. Kllin. Wschr. 10:753-755, 1897.
Stocker, HH. and H.T. Enterine: "Cardiovocal syndrome." Laryngeal paralysis in intrinsic heart disease." Amer. Heart J., 56:51-59, 1958.

PAGET-SCHRÖTTER SYNDROME

Synonymy

S. of traumatic thrombosis of the axillary vein. S. of intermittent venous claudication. S. de Schrötter.

Syndromography

Clinic

It occurs in active, healthy men between 18 and 40 years old.

Symptoms appear after vigorous work or occasionally without exertion or after minimal exertion. Its onset is gradual or very rapid. The right arm is most frequently affected. Increase in volume of the entire limb, from the fingers to the shoulder girdle and lower part of the neck, with dull pain in the joints of the limb, especially the shoulder, sometimes there is no pain. There are no general manifestations.

Physical examination. Increased volume of the arm, diffuse or patchy cyanosis, prominent superficial veins, occasionally an elongated induration is palpated in the axilla.

Paraclinical examinations

Special tests. Venous pressure, oxygen saturation and circulation time show stasis.

Chest radiology. Negative.

Blood. Normal.

Venography. Shows obstruction and collateral circulation.

Syndromogenesis and etiology

Idiopathic obstruction of the axillary-subclavian veins (not caused by aortic aneurysm, heart failure or breast neoplasia); It is probably due to compression of the vein between the muscles and the first costal arch or the clavicle.

It can be traumatic.

Bibliography

Kieny, R.; R. Fontaine: A. Suhler, et al.: Thirty-four cases of so-called exsertion" "Thrombophlebitis of upper extremity (Paget- -Schrötter syndrome)". J. Cardiovasc. Surg., 13:181-185, 1964.
Paget, Sir J.: Clinical Lectures and Essay. London, 1875, p.292.
Schrötter, L.: “Von Erkrankunger der Gefasse in Noth-agels Hand-buch der Pathologie und Therapie.” Wein, 1884. Holder (Noth-nagel), 1884.
Steinberg, I: “Angiographic features of primary venous obstruction of upper extremity: Paget-von Schrötter syndrome". Am.J. Roentgenol., 98:388, 1966.

IDIOPATHIC PERICARDITIS SYNDROME

Synonymy

S. of idiopathic benign pericarditis. S. de Porter. S. of acute benign pericarditis. S. of benign pericarditis. S. of acute recurrent benign pericarditis.

Syndromography

Clínic

It is observed at all ages although it predominates in young men. It occurs 1-2 weeks after an upper respiratory infection. Acute chest pain with wide radiation

especially to the neck and shoulders. It becomes more acute with deep inspiration, coughing, and sometimes swallowing. Reclining increases pain, while sitting decreases it. There is cough, dyspnea, fever.

Physical examination. Pericardial rub, enlargement of the cardiac area. Pleurisy with effusion and pneumonitis are frequently associated.

Paraclinical examinations

Blood. Moderate leukocytosis. Erythrocyte sedimentation rate is rarely accelerated.

Electrocardiogram. There are no destructive changes, sometimes it simulates an acute myocardial infarction.

Pericardiocentesis. It allows obtaining and studying the characteristics of the fluid contained in the pericardial cavity.

Chest radiology.

Syndromogenesis and etiology

The relationship of this syndrome with a viral infection has been proven only in some cases. An autoimmune reaction has also been considered as a possible mechanism. In some cases, gout has been observed and has been considered in the etiology.

Recurrences are frequent.

Literature

Friedberg, CK.: Discases of the Heart. ed. 3th, Philadelphia, Saunders, 196.

Hodges, R.M.: Idiopathic pericarditis". Boston Med. Surg. J., 51:140-141, 1854.

Porter, W.B.; 0. Clark and R.R. Poter: Non-specific benign pericarditis." J.A. M.A., 144:749-753, 1950.

SYNDROME OF PERSISTENCE OF THE DUCT ARTERIOSUS WITH REVERSED FLOW0

Syndromography

Clínic

Sometimes from birth and sometimes during development, dyspnea on exertion occurs, occasionally in the squatting position.

Physical examination. Cyanosis that increases with exercise, more intense in the lower half of the body and in the left hand more than the right. Clubbing toes. Heart: weak systolic murmur in the left third space or absence of murmur. Rarely a strong diastolic murmur at the left edge of the sternum, with thrill.

Paraclinical examinations

Electrocardiogram. Right hypertrophy.

Chest radiology. Prominence of the middle arch and branches of the pulmonary artery, thinning of small peripheral arteries.

Echocardiogram. Overload and dilation of cavities right.

Syndromogenesis and etiology

It is due to the presence of a large, persistent ductus arteriosus with reversal of flow from right to left due to pulmonary hypertension.

Literature

Friedberg, C.K.: Diseases of the Heart. ed. 3, Philadelphia, Saunders, 196.

PATENT DUCT ARTERIOSUS SYNDROME

Syndromography

Clinic

Common in premature babies and individuals born at high altitudes. The child may develop normally, but presents dyspnea on exertion, fatigue, and palpitations. Occasionally, development may be delayed. Sooner or later there is heart failure. Bacterial endocarditis can complicate this syndrome.

Physical examination. Paleness, graceful habit. Heart: Gibson murmur (noise in machinery that occupies systole and diastole) with characteristic increasing systolic thrill at the site of greatest intensity of the murmur; accentuation of the second lung sound;
Corrigan's occasional strong pulse, capillary pulse, gunshot femoral pulse.

Paraclinical examinations

Electrocardiogram. Left ventricular hypertrophy and/or right.

Chest radiology. Increased pulmonary circulation, enlargement of the trunk of the pulmonary artery, enlargement of the left cavities and the aorta, sometimes calcification of the duct is observed.

Echocardiogram. Overload of the volume of the left ventricle, dilation of the left chambers and the aorta.

Syndromogenesis and etiology

Persistence of the ductus arteriosus after 8 weeks after birth. If the defect is small, there is a shunt from left to right and the pulmonary artery pressure remains normal; When the defect is greater, the pressure from the aorta is transmitted to the pulmonary pressure, pulmonary hypertension can be established and the shunt is reversed.

Literature

Friedberg, C.K.: Diseases of the heart. ed. 3th, Philadelphia, Saunders, 1966.

PICK SYNDROME, F.

Synonymy

S. of constrictive pericarditis. Friedel-Pick pseudohepatic cirrhosis. S. of mediastinopericarditis. S for pseudocirrhosis of the liver. S. de Hutinel-Pick. S. of polyserositis. S. of mediastinum-pericardial pseudocirrhosis of the liver.

Syndromography

Clínic

It occurs in patients with a history of chest infection. There is dyspnea, weakness, precordial discomfort, anorexia.

This syndrome is characterized by effusions in the serous membranes, with the symptomatic process specific to each one of them.

There is a chronic, adhesive, constrictive pericarditis and a chronic peritonitis that includes injury to both visceral and parietal serosal tissue.

Physical examination. Jugular engorgement, edema in lower limbs, normal heart or with muffled sounds, paradoxical pulse, protodiastolic gallop, hepatomegaly, ascites, low blood pressure.

There is a variant known as Gouley syndrome or pulmonary artery constrictive pericarditis syndrome, in which this artery is partially or completely compressed, causing cardiovascular symptoms in relation to the injury.

Paraclinical examinations

They depend on the serous effusion or effusions present.

Urine. Normal.

Blood. Low albumin, high bromosulfthalein retention.

Special tests. Increased venous pressure; prolonged circulation time.

Radiology. Calcifications in the pericardium, decreased heartbeat on fluoroscopy; frequently pulmonary and pleural alterations.

Electrocardiogram. Flattening or inversion of T.

Echocardiogram.

Cardiac catheterization.

Punctures and/or Biopsies.

Radiology and especially punctures and/or biopsies are of great value, which serve to determine the etiology.

Syndromogenesis and etiology

The fundamental lesion that causes this syndrome is a chronic pericarditis that in its evolution creates adhesions between the two sheets of the pericardium, which compresses or imprisons the heart, preventing diastole from taking place satisfactorily. The causes that can cause this injury are: tuberculosis, other infections (bacterial or viral), idiopathic.

It is considered that Pick's syndrome is more common in patients who present tuberculous pericarditis, rheumatic fever, ca or pneumococcal infection.

Literature

Madaras, J.S.; RE. Taber and C.R. Lam: "Constrictive pericarditis: diagnosis and operative management". Dis. Chest, 52:746-753, 1967.

Pick, F.: Ueber chronische, unter den Bilde der Lebercirrhose velaufende Pericarditis (pericarditische Pseudolebercirrhose) nebst Bemerkungen uber die Zuckergussleber (Curschmann)".
Zeits. f. Klin. Med., 29:385-4 10, 1896.

Stadler, H. and D. Stinger: "Case of Pick's Syndrome as basis for study of hyproteinemia." J. Pediat., 18:84, 1941.

DIVING FOOT SYNDROME

Synonymy

S. of the foot of the trench. S. of the foot of swamp. S. of the foot of manigua. S. of Paddy's foot. S. tropical dip foot. (See pernio syndrome.)

Syndromography

Clinic

It occurs in soldiers, hunters, individuals who suffer prolonged exposure of the lower limbs to cold and humidity and with little movement.

Physical examination. 1. Initial vasospastic, ischemic phase: edema, paleness, cyanosis of the feet and then petechiae, increased volume and ulcerations. Patch anesthesia. 2. Post-immersion, hyperemic phase. After cessation of exposure to cold, the feet are warm and red, the peripheral pulses full (as in erythromelalgia). Blisters with hemorrhagic fluid. Ecchymosis, ulcerations, gangrene. A burning sensation appears on the tenth day. The condition ends in 2 weeks if gangrene has not occurred. 3. Late vasospastic phase, ischemic phase (in half of the cases well-treated patients do not reach this stage). Coldness, pain, stiffness and paresthesia in the limbs. Exposure to cold triggers Raynaud's phenomenon and hyperhidrosis.

Syndromogenesis and etiology

This syndrome is due to changes in peripheral circulation caused by prolonged exposure to cold and humidity.

Literature

Taplin, D., N. Zaias and N. H. Blank: The role of temperature in tropical immersion foot syndrome." J.A.M.A., 202:546-549, 1967.

Whayne, T.F. and M.E. DeBakey: Cold Injury Ground Type in World War II. Med. Dep. U.S. Army, 1958.

Wright, I.S. and E. V. Allen: "Frostbite, immersion foot, and allied conditions." Army Med. Bull., 65:136-150, 1943.

POST-CARDIAC SURGERY SYNDROME

Synonymy

S. postpericardiotomy. S. postcommissurotomy or cardiotomy.

S. of postperfusion pulmonary congestion.

Syndromography

Clinic

Weeks or months after cardiac surgery: chest pain, fever, myoarticular pain, cough, pericardial friction, dyspnea.

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation. Leukocytosis.

Radiology. Pericardial effusion infiltrated into lung bases, pleural effusion.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Ellis, K., et al.: "Rocntgenographic findings after pericardial surgery", Radiol. Clin. North Am., 9:327, 1971.

Soloff, L.A., et al.: “Reactivation of rheumatic fever following mitral commissurotomy.” Circulation, 8:481, 1953.

POSCOARTATION SYNDROME

Synonymy

S. of postcoartectomy. S. of mesenteric arteritis.

Syndromography

Clinic

Secondary to surgery to resect the coarctated segment of the aorta. On the second day after surgery: anorexia, increased temperature. On the fourth day: abdominal pain and vomiting, abdominal distention.

Physical examination. From the eighth day: paralytic ileus, moderate signs of peritoneal irritation.

Examination demonstrates diffuse petechial stippling in the small intestine. Late hypertension (tenth day).

Paraclinical examinations

Radiology. Paralytic ileus with edema of the intestinal wall, strictures at scar sites as a late sequelae.

Blood. Leukocytosis.

Syndromogenesis and etiology

Excision of the narrowed area exposes the vascular bed beneath the coarctation to increased pulse pressure and constant, large distension of the arteries.

Vasospasm and ischemia of the intestine.

Literature

Ho, E.C.K., et al.: The syndrome of mesenteric arteritis' following surgical repair of aortic coarctation: Report of nine cases and review of the literature", Pediatrics, 49:40, 1972.

Mays, E.T., and C.K. Sergeant: "Postcoarctectomy syndrome" Arch. Surg. 91:58-66, 1965.

POSTPHLEBITIC SYNDROME

Synonymy

Post-thrombotic S. Postphlebitic S. of the legs. S. of venous ulcer. S. of varicose ulcer. S. of chronic venous insufficiency.

Syndromography

Clinic

It appears as a sequel to a venous thrombosis suffered 3-10 years previously. It affects middle-aged women more in the lower limbs. It is bilateral in 25% of cases.

Physical examination. Increase in volume, edema, pain when walking, spots on the skin, sometimes varicose veins and ulcerations.

Paraclinical examinations

Serial phlebographs.

Trendelenburg test.

Perthes test.

venous pressure.

Syndromogenesis and etiology

The syndrome is the consequence of venous hypertension, caused by postphlebitic incompetence of the communicating veins.

Literature

Blalock, A.: "Oxygen content of blood in patients with varicose veins." Arch. Surg., 19:898-905, 1929.

Negus, D.: "Calf pain in the post-thrombotic syndrome". Bri. Med. J.. 2:156-158, 1968.

Nielubowicz, J. and M. Szosteck M.: "The etiology of postphlebitic syndrome." J. Cardiovasc. Surg., 13:140-150, 1964.

POSTPERFUSION SYNDROME

Synonymy

Postcardiotomy febrile lymphocytic splenomegaly.

Syndromography

Clinic

It occurs in young patients undergoing heart surgery who have undergone the cardiopulmonary bypass technique.

Symptoms begin 2-4 weeks after the intervention and are characterized by fever in the absence of chest pain or definite discomfort.

Physical examination. Splenomegaly (70%), transient exudative pharyngitis (5%), adenopathy (20%), hepatomegaly (30%).

Paraclinical examinations

Blood. Negative blood cultures, lymphocytosis (60%) with atypical cells (virocytes). Transaminases and lactic dehydrogenase moderately elevated transiently. Abnormal liver function tests. Heterophile antibodies: negative or moderately elevated. Anemia and hyperbilirubinemia in some cases.

Syndromogenesis and etiology

Possible viral infection due to its apparent seasonal distribution. It has been considered that it is a leukocyte graft against host reaction (?).

The etiology is unknown.

Bibliography

Kahn, D.R.; PH. Ertel; W.H. Murphy, et al.: "Pathogenesis of the postcardiotomy syndrome." J. Thorac. Cardiov. Surg., 54:682- -687, 1967.

Kreel, I.; L.I. Zaroff; J.W. Carter, et al.: “A syndrome following total body perfusion." Surg. Gynec. Obstet., 111:317-321, 1961.

Reyman, TA.: "Postperfusion syndrome." Amer. Heart J. 72:116-123, 1966.

POSPERICARDIOTOMY SYNDROME

Synonymy

S. postcommissurotomy. S. postcardiotomy (see Dressler syndrome). S. postvalvotomy.

Syndromography

Clínic

Postoperative complication that appears weeks or months after an intervention on the heart. The patient may present slight general disorders. Pleuropericardial pain, sometimes stabbing, radiating to the epigastrium, shoulders and back, aggravated by changes in position and deep inspiration: fever, occasional cough, occasional muscle and joint pain.

Physical examination. Pericardial rub, signs of pericardial and pleural effusion (unilateral or bilateral). Signs of congestive heart failure. There are no peripheral venous thromboses or pulmonary infarction. There is polyarthritis, subcutaneous nodules.

Paraclinical examinations

Chest radiology.

Electrocardiogram.

Blood. Accelerated erythrocyte sedimentation rate, leukocytosis with neutrophilia, occasionally eosinophilia and anemia.

Syndromogenesis and etiology

Various hypotheses have been proposed to try to explain the symptoms of the syndrome: possible response to surgical trauma, consequence of the incision of the pericardium, release of antigens and hypersensitivity reaction.

The etiology is unknown.

Literature

Soloff, L.A.: J. Zatuchni: 0.H. Janto, et al.: "Reactivation of rheumatic fever following mitral commissurotomy." Circulation, 8:481-493, 1953.

PRINZMETAL OR ANGINOID SYNDROME

Synonymy

Prinzmetal's variant of angina (1959). S. of reversible angor pectoris. Variant angina pectoris. S. of reverse angina pectoris.

Syndromography

Clinic

Anginal pain occurs, but unlike classic angina pectoris, the pain appears more frequently at rest, is longer lasting and more intense, and is not related to effort nor does it subside with rest, since it is not due to an increase in cardiac work. It usually appears at the same time of day and may be accompanied by syncope.

Paraclinical examinations

Electrocardiogram. ST segment elevation as occurs in acute transmural myocardial infarction, instead of depression during the painful crisis. Transient ST segment elevation is accompanied by discordant ST segment depression in other leads. Transient AV block in some patients.

Syndromogenesis and etiology

This syndrome is considered to be caused by temporary occlusion of a coronary artery with eventual development of an infarction at the location of the myocardium, indicated by transient electrocardiographic changes.

Other mechanisms have also been considered such as:

coronary arterial spasm due to increased adrenergic tone, anomalous branching of the coronary arteries.

Literature

Prinzmetal, M.; A. Ekmekei; R. Kennamer et al.: "Variant form of angina pectoris, previously undelineated syndrome." J.A.M.A., 174:1794-1800, 1960.

MITRAL VALVE PROLAPSE SYNDROME

Synonymy

S. of prolapse of the posterior mitral leaflet. S. of aneurysmal protrusion of the posterior leaflet of the mitral valve.

S. for clicky puff. S. mitral valve flutter. S. of mid-systolic click.

Syndromography

Clinic

This syndrome occurs in both sexes, young adults, but is more common in women and occasionally in families. It is generally asymptomatic, but there may be atypical precordial pain reminiscent of angina pectoris, palpitations. Rarely, there are symptoms of left heart failure, endocarditis, progressive mitral regurgitation.

siva or sudden death.

Sometimes it has a vague psychoneurotic basis, hyperventilation

Physical examination. Absence of objective findings in relation to symptoms, except paroxysmal arrhythmias and auscultation that is characterized by the auscultation of a mid-systolic click accompanied by a late systolic murmur, which gives the syndrome its name. Sometimes orthostatic hypotension.

Paraclinical examinations

Electrocardiogram. Almost always normal; occasionally ST depression and T wave inversion in DI-DIL. Prolonged QT, extrasystoles, paroxysmal tachycardia.

Phonocardiogram. Mid-systolic click (prerequisite) and often end-systolic murmur.

Chest radiology. Almost always normal if there is no significant mitral regurgitation.

Echocardiogram. Systolic protrusion of the anterior, posterior, or both mitral leaflets into the left atrium.

Angiography. Ballooning of the posterior leaflet, mitral regurgitation, abnormal convexity of the lower edge of the left ventricle during systole.

Syndromogenesis and etiology

It is common in Marfan syndrome and other connective tissue diseases. Its incidence is increased in patients with atrial septal defect (ostium secundum) and hypertrophic cardiomyopathy.

Prolapse of the mitral valve or of a chorda tendineae or other mitral valve disease can cause it.

Chest pain may be due to ischemia or tension of the papillary muscles.

The etiology corresponds to connective tissue defects and, in addition, it has a familial tendency and autosomal dominant inheritance.

Literature

Barlow, J.B. and C.K. Bosman: “Aneurysmal protrusion of the posterior rior leaflet of mitral valve, an auscultatory-electrocardiographic syndrome'". Amer. Heart J., 71:166-178, 1966.

Hancock, E.W. and K. Cohn: The syndrome associated with midsystolic click and late systolic murmur". Amer. J. Med., 41:183- -196, 1966.

Humphries, J.O. and V. A. McKusick: The differentiation of organic and "innocent' systolic murmurs". Program Cardiovasc. Dis., 5:152-171, 1962.

Jeresaty, R.M.: “Mitral valve prolapse-click syndrome.” Program Cardiovasc. Dis., 15:623, 1973.
Reid, J.V.: Midsystolic clicks." South Afican Med. J, 35:353-355, 1961.
Rizzon, P. et al.: “Familial syndrome of midsystolic click systolic murmur.” Br Heart J., 35:245. 1973.

PSEUDONFARTCH SYNDROME DUE TO ACUTE MYOCARDITIS

Synonymy

Pseudoepicardial S.

Syndromography

Clínica

1. Violent abdominal pain.
2. Early circulatory collapse.
3. Absence of fever.

Paraclinical examinations

Electrocardiogram. Low voltage and signs suggesting previous infarction.

Syndromogenesis and etiology

This syndrome can be caused by: fatal subacute myocarditis preceded a week earlier by a nonspecific infection, by an inflammatory or toxic lesion, or by circulatory disorders.

SILENT LUNG SYNDROME IN CONGESTIVE HEART FAILURE

Syndromography

Clinic

It occurs in the period of incomplete recovery from congestive heart failure. There is mild dyspnea and fatigue.

Physical examination. Clear lungs, no obvious signs of heart failure (edema, hepatomegaly).

The presence of hepatojugular reflux with prolongation of arm-tongue circulation time reveals the persistence of a significant degree of congestive heart failure.

Syndromogenesis and etiology

It is common in any congestive heart failure when right failure is secondary to left failure and the patient has been treated. In these cases an apparent compensation is achieved.

Literature

Schwartz, M.J.: "The 'silent' lung in biventricular heart failure." Amer. J. Cardiol., 6:1032-1035. 1960.

RAYNAUD SYNDROME

Synonymy

Raynaud's disease? Raynaud's phenomenon secondary.

Syndromography

Clínica

Raynaud's disease, which is also known as idiopathic Raynaud's syndrome, must be differentiated from Raynaud's phenomenon syndrome. We will describe the idiopathic type.

It is more common in females (5:1), generally beginning in the second or third decade of life. As a rule, cold is the precipitating factor that reveals the clinical picture. Stress is another precipitating factor involved.

In the fingers there are episodes of color changes at the beginning and gradual increase, occasionally it has a dramatic beginning with intense manifestations (dead finger phenomenon). Generally, after exposure to cold, pallor, cyanosis and redness appear on the fingers, followed by pain and paresthesia. Initially, only the tips of one or two fingers are grasped. Over time, the phenomenon spreads to all the fingers and the rest of the hand. It can appear on the feet. Bilaterality is a diagnostic criterion, since Raynaud's phenomenon is the one that occurs with unilateral location.

Absence of systemic manifestations, which are typical of Raynaud's phenomenon.

Physical examination. Transient color changes in the fingers, coldness and slight swelling during attacks. Sclerodemic changes in the skin after several attacks and, occasionally, small gangrenous lesions.

Paraclinical examinations

Special tests. All diseases associated with Raynaud's phenomenon must be ruled out (see this one).

Syndromogenesis and etiology

It is a vasospastic functional condition. Raynaud's syndrome (idiopathic) must be differentiated from Raynaud's phenomenon, thromboangiitis obliterans, arteriosclerosis obliterans, Weir Mitchell I syndrome and scleroderma or systemic sclerosis.

Literature

Raynaud, A.G.M.: De l'aphyxie local ct de la gengrene symetrique des extrémites. Paris, Rignoux, 1862, pp.6-9.

CORONARY ARTERY STEAL SYNDROME

Synonymy

S. of intramural aneurysm of the coronary artery. S. of the last coronary artery that emerges from the pulmonary artery.

Syndromography

Clinic

It appears in childhood shortly after developing adequate collateral circulation, or in adults, in cases with abnormal origin of the left coronary artery in the pulmonary artery (Bland-White-Garland syndrome). Also in childhood, whenever a communication between the coronary arteries and the cardiac chambers or an intramural coronary aneurysm steals blood from the coronary circulation. It is characterized by anginal precordial pain, dyspnea, fatigue on exertion, sweating, paleness, crying after exertion or ingestion of food.

Physical examination. Harsh systodiastolic murmur along the left edge of the sternum and at the tip, or machine murmur with reinforcement of the second lung sound.

Cardiomegaly at the expense of the left ventricle, progressive heart failure.

Paraclinical examinations

Electrocardiogram. Inverted T wave in DI-DII, deep Q wave in DI; signs of anterolateral infarction.

Angiography of the left ventricle and aorta. Normal left ventricle and right coronary artery; descending branch of the left coronary artery dilated 1 cm, tortuous, emptying into an aneurysmal sac at the tip of the heart; The aneurysm dilates in diastole and contracts in systole.

This test is diagnostic.

Chest radiology. Cardiomegaly with left predominance. Sometimes pulmonary edema.

Syndromogenesis and etiology

It is a congenital anomaly of the left coronary artery that arises in the pulmonary artery, or an intramural aneurysm of the coronary artery.

Literature

Baue, A.E.; S. Baum; W.S. Blakemore, et al.: A later of anomalous coronary circulation with origin of the left coronary artery from the pulmonary artery." Circulation, 36:878-888, 1967.

Bjork, V.0. and L. Bjork: "Intramural coronary artery aneurysm. A coronary artery syndrome". J. Thor. Cardiovas. Surg., 54:50- -52, 1967.

Wesselhoeft, H., et al.: “Anomalous origin of the left coronary artery from the pulmonary trunk: Its clinica.” spectrum, pathology, and pathophysiology, based on a review of 140 cases with seven further cases. Circulation, 38:403, 1968.

SUBCLAVIA ROB SYNDROME

Synonymy

Congenital pulmonary-subclavian steal. S. of brachial-basilar deficiency. S. of basilar arterial insufficiency.

Syndromography

Clinic

It is characterized by lack of irrigation in the affected arm with increased fatigue during movements. It is usually more common in the left arm, but can be bilateral.

It is frequently accompanied by: numbness, coldness, claudication and pain in the arms and hands. Exercise with the affected limb precipitates neurological symptoms: syncope, facial paresthesia, loss of vision and headache.

Sometimes there is cyanosis and signs of respiratory failure and also signs of cerebral vascular insufficiency, with loss of consciousness and even seizures.

Physical examination. Difference in pulse amplitude in both arms, as well as in blood pressure, with evident reduction in humeral pulse and blood pressure in the ipsilateral limb. Absence of carotid pulse on that side.

Murmur under the supraclavicular area. On cardiac auscultation: intense systolic murmur (rarely diastolic) to the left of the sternum.

Paraclinical examinations

Radiology. Attenuation of the vascular network of the lung.

Cardiomegaly. Aorta on the right side.

Arteriography. It is essential to specify arterial variations. Retrograde or decreased blood flow in the vertebral artery.

Syndromogenesis and etiology

The symptoms occur because when there is a stenosis of the proximal portion of the subclavian artery, blood drains from the vertebral artery to the upper limb, which causes basilar insufficiency, that is, it flows in the opposite direction through the right carotid artery, due to occlusion of the innominate artery (brachiocephalic trunk). There are three varieties according to the emergence of the subclavian artery and the brachiocephalic trunk.

The etiology is due to congenital or acquired atherosclerosis: thrombosis, extrinsic compression or stenosis of the subclavian artery proximal to the vertebral branch.

Literature

Berger, R.L.: JJ. Sidd and K. Ramaswamy: Retrograde vertebral-artery flow produced by correction of subclavian-steal syndrome." New Engl. J. Med., 277:64-69, 1967.

Contorni, L.: "I circolo collaterale nella legatura dellárteria succlavia." Minerva Chir.. 15:268-271, 1960.

Massumi, R.A.: "The congenital variety of the 'subclavian steal' syndrome." Circulation, 28:1149-1152, 1963.

Perloff, J.K.: Neurological disorders and heart disease. Heart diseases. (2nd ed.) Philadelphia. Saunders, 1984.

SCHROEDER SYNDROME

Synonymy

Acute S. of salt depletion. S. of hyponatremia hypotonic hypovolemia. S. of acute salt loss.

Syndromography

Clinic

During intense treatment of congestive heart failure, rapid physical and mental deterioration is seen due to the indiscriminate use of powerful diuretics (formerly mercurial). Thirst, anorexia, nausea, prostration, cramps in the calves, tendency to syncope in the standing position, oliguria and, finally, kidney failure.

Paraclinical examinations

Blood. Predominantly chlorine or sodium depletion as well as chlorine. Hyperazoemia. Hemoconcentration.

Syndromogenesis and etiology

The syndrome is caused by acute dehydration with loss of chloride and sodium after iatrogenic profuse diuresis. It should not be confused with chronic dilutional hyponatremia syndrome (see this one).

Literature

Schroeder. H.A.: "Renal failure associated with low extracellular sodium chloride: the low salt syndrome." J.A.M.A., 141:117--124, 1949.

Vogl, A.: "The low-salt syndromes in congestive heart failure." Amer. J. Cardiol., 3:192-198, 1959.

SHY-DRAGER SYNDROME

Synonymy

Shy-Drager's idiopathic orthostatic hypotension syndrome. Shy-Drager variant orthostatic hypotension.

S. of idiopathic autonomic failure.

Syndromography

Clinic

It occurs in adults. The onset is gradual: orthostatic hypotension, impotence, generalized or localized anhidrosis (Bradbury-Eggleston triad). Sphincter disorders, visual disorders, rigidity, tremor, adiadochokinesia, fasciculations, muscle exhaustion that can lead to disability. Occasionally dysphagia. The absence of sensory changes and mental deterioration is striking. There is no compensatory tachycardia.

Physical examination. Iris atrophy, ophthalmoplegia, osteotendinous hyperreflexia.

Paraclinical examinations

Blood. Only electrolyte disorders.

Electroencephalogram.

Electromyography.

Urine. Steroid excretion.

Acetylmethylcholine injection. Absence of response.

Syndromogenesis and etiology

Progressive multisystem degeneration of the nervous system. The inflammatory nature cannot be excluded. A slowly evolving familial form has been described.

The etiology is unknown.

Literature

Bradbury, S. andC. Eggleston: Postural hypotension: Report of three cases." Amer. Heart J., 1:73-86, 1925.

Shy, GM. and G.A. Drager: "A neurological syndrome associated with orthostatic hypotension". Arch. Neurol., 2:511-527, 1960.

Sparberg, M., K.B. Kundsen and S.T. Frank: Dysautonomia and dysphagia." Neurology, 18:504-506, 1968.

STOKES-ADAMS SYNDROME

Synonymy

S. Adams-Stokes. S. de Morgagni-Adams-Stokes. S. of Spen. S. of complete heart block.

Syndromography

Clínica

Syncope or convulsions preceded by a feeling of malaise, dizziness, dizziness, chest tightness.

Physical examination. Extreme bradycardia, arterial hypotension, seizures, loss of consciousness of very short duration.

Paraclinical examinations

Electrocardiogram. Bradycardia due to complete atrioventricular block. Sometimes atrial fibrillation

asystole.

Syndromogenesis and etiology

Cerebral hypoxia due to temporary cardiac arrest determined by the block.

This phenomenon can be triggered by: extracardiac nervous influences, which may have their origin in the medulla, circulatory center or vagus nerve; psychic stimuli.

The anatomical substrate is constituted by a toxicodegenerative inflammatory alteration or a congenital malformation of the myocardium at the level of the interventricular septum that produces an aplasia of the Kent fasciculus.

The most accepted mechanism is the sudden reduction in cardiac output and blood pressure, which determines cerebral hypoxia, which is due to cardiac arrest caused by the blockage.

Although the classic one is described by atrioventricular block in the so-called paralytic Stokes-Adams syndrome, it can also occur during the course of other tachyarrhythmias (paroxysmal supraventricular tachycardia), it is the so-called dynamic Adams-Stokes syndrome and others such as sinus-atrial block.

It is considered that loss of consciousness in Stokes-Adams due to atrioventricular block may be caused by: 1, absence of an idioventricular escape rhythm or delay in its initiation; 2, asystole during the transition from sinus rhythm to complete block; 3, occurrence of ventricular arrhythmia or fibrillation.

Literature

Adams, R.: “Cases of diseases of the heart, accompanied with pathological observations.” Dublin Hosp. Rep., 4:353, 1827.

Morgani, G.B.: De sedibus et causis morborum. Lib. 1, p. 70.1761.

Stoke, W.:*Observations on some cases of permanently slow pulse" Dublin Quart. J. Med. Sc., 2:73-85, 1846.

TAUSSIG-BING SYNDROME

Synonymy

S. of the biventricular pulmonary artery-vascular transposition. S. of the Taussig-Bing complex. S. of the double outlet of the right ventricle (Neufeld type IIa).

Syndromography

Clinic

Dyspnea on exertion and cyanosis from birth.

Physical examination. Cyanosis, cardiomegaly and insignificant systolic murmur in the left third intercostal space, reinforcement of the second lung sound, clubbing of the fingers, terminal heart failure. Delay in development.

Paraclinical examinations

Electrocardiogram. Right ventricular hypertrophy with right axis deviation.

Chest radiology. Moderate to great cardiomegaly, increased pulmonary circulation.

Angiocardiography. Simultaneous opacification of the aorta and pulmonary starting from the right ventricle, the aorta being denser than the pulmonary one. In the lateral view, the aorta is located in front of the pulmonary artery.

Blood. Polycythemia, moderate.

Syndromogenesis and etiology

It is a congenital malformation in which the aorta leaves the right ventricle and the pulmonary artery rides on the septum due to a high septal defect.

Literature

Neufeld, H.N., et al.: "Origin of both great vessels from the right ventricle: I. Without pulmonary stenosis. II. With pulmonary stenosis." Circulation, 23:399, 1961.

Taussig, H.B. and R.J. Bing: "Complete transposition of aorta and levoposition of pulmonary artery." Amer. Heart J., 37:55 1-559, 1949.

What is the Taussig-Bing malformation? (Editorial), Circulation, 38:445-49, 1968.

TETRALOGY OF FALLOT SYNDROME

Synonymy

S. of blue evil. S. of coeruleo morbidity.

Syndromography

Clínica

In the first weeks of life, the child has difficulty feeding, poor body-stature development, dyspnea or fatigue on exertion, orthostatic dyspnea. After a few months of life, paroxysms of hypoxic hyperpnea with increased cyanosis (alarming manifestations), which disappear before the age of 6 years. The patient crouches (squats) when tired or dyspnea to improve symptoms (Taussig position).

Physical examination. Cyanosis since childhood, at first only during crying or exercise and then persistent. Clubbing fingers on hands and feet. Systolic murmur at the level of spaces II and III next to the left edge of the sternum, with thrill. Single second sound due to the absence of the pulmonary element. A systolic beat can be observed in spaces II and II of the midclavicular line.

Paraclinical examinations

Radiology. Elevation of the tip of the heart, prominent left upper arch, decreased pulmonary circulation, absence of pulsation of the hilar pulmonary vessels.

Angiocardiography. Informs about the location of the defect.

Electrocardiogram. Right axial deviation, depressed ST, inverted T in DII-DIIL.

Cardiac catheterization. It reveals increased pressure in the right ventricle, decreased pressure in the pulmonary artery and altered oxygen pressure in all cavities. Arm-tongue and right ventricle-ear circulation time greatly shortened.

Blood. Polyglobulia.

Syndromogenesis and etiology

The syndrome is caused by a congenital heart disease with multiple defects: ventricular septal defect, pulmonary stenosis, dextroposition of the aorta, ventricular hypertrophy. Trilogy of Fallot syndrome only presents: pulmonary stenosis, atrial septal defect, right ventricular hypertrophy.

Bibliography

Fallot, A.: "Contribution á l'anatomie pathologique de la maladie bleue (cyanose cardiacaque)". Marseille Méd., 25:77, 138, 207- -210, 341, 403, 1888.

Johnson, C.: Fallot's tetralogy: A review of the radiological appearances in thirty-three cases". Clin. Radiol., 16:199, 1965.

Stayanaryana, R., et al.: "Anatomic variations in the tetralogy of Fallot." Am. Heart J., 81:361, 1971.

TROUSSEAU SYNDROME

Synonymy

S. of migratory thrombophlebitis. S. of carcinogenic thrombophlebitis. S. of idiopathic recurrent thrombophlebitis. S. of superficial venous thrombosis.

Syndromography

Clínic

The lower limbs are the most frequently affected; When located in the upper limbs it has greater diagnostic value. It is characterized by repeated episodes of pain that persist for months or years.

Physical examination. Prominent, erythematous, tender, rope-like subcutaneous nodules that disappear without leaving a trace or very little damage before new lesions appear.

Paraclinical examinations

Special tests. The fundamentals for the search for a hidden malignant process.

Syndromogenesis and etiology

Frequent association with carcinoma, especially of the pancreas; Possible release by the neoplasm of a substance similar to thromboplastin. Neoplasms related to recurrent migratory phlebitis tend to be inoperable.

The etiology is unknown.

Literature

Quick, A.J.: Hemorrhagic Diseases and Thrombosis. ed. 2, Philadelphia, Lea & Febiger, 1966.

Thompson, A.V.: Thrombosis of visceral veins in visceral cancer." Clin. J., 67:137-140, 1938.

Trousseau, A.: Lectures delivered in 1862.

CAROTID CORPUSCLE TUMOR SYNDROME

Synonymy

S. of non-chromaffin paraganglioma of the carotid corpuscle.

Syndromography

Clínic

This syndrome occurs with the same frequency in both sexes, appearing from the second to fourth decades of life, on average around 45 years of age. When the diagnosis is made, the symptoms last about 2 years. It is frequently asymptomatic, it can manifest itself as hoarseness, vertigo, facial paralysis, and vocal cord paralysis.

Physical examination. A cervical mass is palpated that moves laterally, but not vertically. Hypopharyngeal mass. The masses are frequently pulsatile, but noises are rarely heard. Metastases are not common.

Paraclinical examinations

Carotid angiography.

Syndromogenesis and etiology

The syndrome is caused by a tumor, non-chromaffin paraganglioma, of the carotid bifurcation.

Literature

Lattes, R.: "Nonchromaffin paraganglioma of ganglion nodosum, carotid body, and aortic-arch bodies." Cancer, 3:667-694, 1950.
Oberman, H.A.; F Holtz; THE. Sheffer et al.: "Chemodectomas (nonchromaffin paraganglioma) of the head and neck." Cancer, 21:838-85 1, 1968.

PARACHUTE VALVE SYNDROME

Synonymy

S. of valvular myxomatous transformation. S. of valvular mucinous degeneration. S. de Read.

Syndromography

Clinic

It is more common in men between 30-60 years old. The symptoms are similar to those of mitral or aortic insufficiency. The insufficiency is usually isolated, but sometimes stigmata of Marfan syndrome can be observed in the patient or their relatives.

Paraclinical examinations

Special tests. Those indicated in valvular insufficiency. Determination of indices for the diagnosis of Marfan syndrome (see this one).

Serology.

Echocardiogram. Prolapse of the valve cusp.

Syndromogenesis and etiology

The mechanisms that cause aortic insufficiency are:

1, reduction in intrinsic valve stiffness whereby the sigmoid leaflets stretch, roll, wrinkle, invert, and prolapse; 2, losses of substances that give rise to a defect in the valve; 3, transformation facilitates infection.

Myxomatous transformation of the leaflets can be observed in incomplete Marfan syndrome, idiopathic dilation of the aorta and accompanying Erdheim necrosis.

The anatomical structure of the myxomatous lesion is given by the appearance of different degrees of hyalinization, disruption and loss of the architecture of the normal connective tissue, with an increase in the intercellular substance and in some cases, fibrosis.

Literature

Frable, W.J.: "Mucinous degeneration of the cardiac valves: The floppy valve' syndrome." J. Thorac. Cardiovasc. Surg., 58:62, 1969.
Read, R.C. and P. Wolf: *Symptomatic valvular myxomatous degeneration (floppy valve syndrome)". Circulation, Suppl. 3, 30:143-144, 1964.
Read, R.C.; AP Thal and V.E. Wendt: "Symptomatic valvular myxomatous transformation (the floppy valve syndrome). A possible forme frustration of the Marfan syndrome." Circulation, 32, 897-910, 1965.
Sherman, E.B., et al.: "Myxomatous transformation of the mitral valve producing mitral insufficiency: Floppy valve syndrome." Am. J. Dis. Child.. 119:171, 1970.

FERIOR VEIN CAVA SYNDROME

Synonymy

S. compression of the inferior vena cava.

Syndromography

Clínic

It is characterized by: edema of the lower limbs, superficial venous engorgement in both lower limbs, abdomen and thorax. Ascites (less common).

Syndromogenesis and etiology

Obstruction or compression of the cava due to thrombosis, neoplasms, aneurysm, adhesions, anomaly, etc.

INFERIOR VENA CAVA OBSTRUCTION SYNDROME

Syndromography

Clínic

The symptoms are related to the location and speed of the obstruction; There is vomiting, diarrhea.

Physical examination. Edema in lower limbs; dilation of the superficial veins of the lower limbs, abdomen and thorax. Occasionally there is ascites.

Paraclinical examinations

Urine. Albuminuria.

Blood. Hyperbilirubinemia. Other liver function tests altered, increased urea nitrogen.

Cavography.

Computerized axial tomography.

Syndromogenesis and etiology

The causes that can cause this syndrome are varied: congenital, aneurysm, thrombosis, abdominal malignancy, infection, trauma, liver cirrhosis.

Literature

Pleasants, J.: "Obstruction of the inferior vena cava with a report of eighteen cases." Johns Hopkins Hosp. Rep., l6:363-558, 1911.

SUPERIOR VEIN CAVA SYNDROME

Synonymy

S. superior mediastinal.

Syndromography

Clinic

It predominates in men of any age with maximum frequency between 40-50 years. It is characterized by venous engorgement

and edema in the face and neck. Dyspnea, orthopnea, dysphagia, hoarseness, epistaxis, headache, vertigo, noises in the ear, drowsiness, syncope.

Physical examination. Edema and cyanosis of the face, neck, shoulders and arms (sclavine edema and cyanosis), conjunctival injection. Collateral circulation in the shoulders and upper chest region. The edema is limited to the indicated regions and has a clear line of demarcation. Edema and cyanosis are also seen in the mucous membranes of the mouth, pharynx, and larynx. There is hydrothorax and, occasionally, hydropericardium.

Paraclinical examinations

Chest radiology. Widened mediastinum with density parallel to the vertebral column (azygos and hemiazygos), hydrothorax, hydropericardium. Dense uniform or rounded or multi-lobed shadows in the upper mediastinum.

Upper cavography. Demonstrates the site of obstruction and collaterals.

Syndromogenesis and etiology

This syndrome can be produced by any cause that compresses, obstructs or infiltrates the superior vena cava or obstructs its circulation: lymphadenopathy, mediastinal neoplasms, aortic aneurysm, cancer of the lung or adjacent organs, intrathoracic goiter and idiopathic causes, radiation, fibrosis, thrombosis, thrombi, mediastinal emphysema, mediastinitis, pericarditis, pulmonary artery anastomosis. right-superior vena

Literature

Brown, R.C., et al.: "Angiographic demonstration of collateral circulation in a patient with superior vena cava syndrome."
Am. J. Roetgenol., 119:543, 1973.

Corvisat,: Essai sur les maladies et les lesions organiques du coeur.
Paris, 1806, p. 350.

Hunter, W., 1757 quoted by B.A. MCArt, FB. Ramsey, W.A. Tosik, et al.: * surgical reversal of superior vena cava syndrome: report of a case caused by intrathoracic goiter and associated with roentgenographic hilar vascular shadow simulating neoplasm of chest". Arch. Surg., 69:4-11, 1954.

Roberts, D.J.Jr. et al.: *Superior vena cava and innominate veins:
Angiographic study." Am. J. Roentgenol., 66:341, 1951.

Vanker, V.P. and FAITH. Maddison: "Superior vena cava syndrome secondary to aortic dissease: report of two cases and review of the literature." Dis. Chest, 51:656-662, 1967.

WOLFF-PARKINSON-WHITE (WPW) SYNDROME

Synonymy

S. of pre-excitation. S. of abnormal atrioventricular excitation. S. of false bundle or branch block. S. of atrioventricular accessory conduction.

Syndromography

Clinic

Generally, it is asymptomatic. The beginning in ages early.

Episodes of paroxysmal tachycardia occur in 10% of patients. cases.

Physical examination. Negative except in tachycardia crises.

Paraclinical examinations

Electrocardiogram. Short PR with elongated QRS (WPW pattern). Type A (positive delta wave in V1 and V6). Type B (negative delta wave in V1 and positive in V6).

Vectocardiogram.

Syndromogenesis and etiology

It is considered a congenital variation of the atrioventricular conduction system due to the presence of an anomalous accessory pathway (Kent's bundle), which results in early excitation of the ventricles.

Literature

Lamb, L.E.: "Wolff-Parkinson-White syndrome." Postgrad. Med., 43:173-176, 1968.

Lind, J.: "Pre-excitation the ventricular part of the heart and its occurrence in children." Acta Paediatr., 32:153, 1945.

Wolff, L.: J. Parkinson and PD. White: Bundle branch block with short P-r interval in healthy young people prone to paroxysmal tachycardia." Amer. Heart J., 5: 685-704, 1930.

RELATED CARDIOVASCULAR SYNDROMES

Syndromes with abdominal angina

Celiac artery.

Superior mesenteric artery.

Mesenteric arteritis.

Abdominal hemicrania.

Median arcuate ligament.

Median aortic obliteration.

Ortner.

Mesenteric artery steal.

Renal artery steal.

Aortoiliac steal (Leriche).

Syndromes with supravalvular aortic stenosis

Uncomplicated supravalvular stenosis.

Complicated supravulvar stenosis.

(See also infantile hypercalcemia syndrome.)

Syndromes with Raynaud's phenomenon

Miscellaneous conditions:

Cold haemagglutination.

Dermatomyositis.

Lupus erythematosus.

Marchiafava-Micheli.

Rheumatoid arthritis.

Scleroderma.

Waldenstrom I.

Neurogenic conditions:

Carpal tunnel.

Diseases of the nervous system.

Compression of the shoulder ring (girdle).

Post-traumatic conditions:

Frosbitc.

Jackhammer.

After the surgical attack.

Vasospasm of pianists and typists.

Occlusive arterial diseases

Arteriosclerosis obliterans.

Embolism.

Thromboangiitis obliterans.

Thrombosis.

Poisoning

Ergotamine.

Heavy metals.

Raynaud.

Syndromes with vascular lesions

Non-relatives:

Fordyce.

Hasabach-Meritt.

Klippel-Trenaunay-Weber.

Maffucci.

Jejunal-oral-scrotal phlebectasia.

Dominant family member:

Blue vesicular-elastic nevus.

Fabry.

Louis-Bars.

Rendu-Osler-Weber.

Sturge-Weber.

Von Hippel-Lindau.

Wyburn-Mason.

Post-heart surgery syndromes

Postcardiotomy diphasic.

Postperfusion pulmonary congestion.

Postcommissurotomy. Postcardiotomy.

Postperfusion.

Postpericardiotomy.

Syndromes with syncope

Temporary loss of consciousness observed in different conditions:

Bouveret.

Hypertensive encephalopathy.

Cerebrovascular diseases.

Aortic stenosis.

Epileptic.

Heberden.

Idiopathic orthostatic hypertensive.

Pulmonary hypertension.

Hyperventilation.

Hysteria.

Migraine.

Myocardial infarction.

Syncopal due to involuntary urination.

Stokes-Adams.

Cough.

Vasovagal.

Weiss-Baker.

Necrotizing vasculitis syndromes

Bazin.

Degos.

Acute febrile neutrophilic dermatosis.

Erythema elevatum duitinun.

Erythema nodosum.

Lethal midline granuloma.

Wegener's granulomatosis.

Horton.

Kussmaul-Mayer.

Lindberg.

Löfgren.

Schönlein-Henoch.

Strauss-Churg-Zak.

Cutaneous-intestinal-oropharyngeal ulcerations.

Urbach.

Allergic vasculitis.

Vilanova-Aguadé.

Whitfeld.

Zeek.

ACHOR-SMITH SYNDROME

Synonymy

S. of nutritional deficiency with hypokalemia.

Syndromography

Clinic

It is a clinical disorder characterized by a marked chronic nutritional deficiency, pernicious macrocytic anemia, diarrhea, steatorrhea (sprue), pellagra (dementia, dermatitis and diarrhea).

Paraclinical examinations

Blood. Hypokalemia, hypochloremia and hypocalcemia.

Syndromogenesis and etiology

This is a chronic multiple nutritional deficiency.

BARLOW SYNDROME

Synonymy

S. de Barlow-Chendle-Moeller. S. of subperiosteal hematoma. S. of infantile scurvy.

Syndromography

Clinic

Discomfort in the legs, pseudoparalysis, irritability, anorexia, diarrhea, gingivorrhagia, hemorrhagic manifestations on the skin (4%), fever (19%), edema over the region of the long bones (14%).

Paraclinical examinations

Blood. There is microcytic, occasionally macrocytic, anemia. Low levels of vitamin C.

Urine. No excretion of vitamin C after a 200 mg IV injection.

Radiology. The bark of the bones appears as a dense line similar to ground glass.

Syndromogenesis and etiology

Barlow syndrome is due to a vitamin C deficiency.

Literature

Barlow, T.: "On cases described as 'acute rickets' which are probably a combination of scurvy and rickets, the scurvy being an essential, and the rickets a variable element". Med. Chir. Trans., 66:159-220, 1883.

Woodruff, C.: “Infantile scurvy, the increasing incidence of scurvy in the Nashville area." J.A.M.A., 161:448-456, 1956.

RIBOFLAVIN DEFICIENCY SYNDROME

Synonymy

S. of arriboflavinosis. Vitamin B2 deficiency.

Syndromography

Clinic

The clinical picture varies according to the intensity of the deficiency. In its intense form, seborrhea, dermatitis in the scrotum, angular stomatitis, cheilosis, fissure of the wing of the nose, pruritus, burning, glossitis, photophobia and abnormal pigmentation of the iris can be observed.

Syndromogenesis and etiology

Dietary deficiency of riboflavin.

CENTRAL NERVOUS SYSTEM DEFICIENCY SYNDROME

Syndromography

Clinic

This is a condition that has been described in prisoners of war and in populations with very low quality nutrition and is characterized by visual deficits that are usually intense accompanied by perceptual hearing loss and tinnitus, gait disorders with instability in maintaining balance.

In the fundus of the eye, paleness of both papillae can be seen.

Syndromogenesis and etiology

The etiology of this condition seems to be nutritional deficiency.

Literature

Garland, H. G.: “A central nervous deficiency syndrome", Proc. Roy. Soc. Med., 39:178, 1946.

GOPALAN SYNDROME

Synonymy

S. with hot feet. S. of electric feet. S. of nutritional melalgia.

Syndromography

Clínic

Clinical picture characterized by sensitivity disorders (intense paresthesias especially in the feet) caused by tingling, electrical sensations, pain that can

be of intense character; sensation of sharp cutting or crushing. Sometimes there may be perceptions of sensitivity induced by tactile stimuli that give rise to aberrant sensations or with anomalous irradiation (localization signs are lost) and that persist when the stimulus ceases. Excessive sweating. Stress taguicardia.

Physical examination. An increase in temperature and vasomotor changes in the feet, walking with tired steps, decreased reflexes, sensory loss, ataxia and weight loss can be observed. Without appropriate treatment this condition can quickly lead to death.

Paraclinical examinations

Blood. Decrease in serum proteins.

Histology. Trophic type changes in the affected areas.

Syndromogenesis and etiology

It is observed in alcoholic beriberi, in the elderly and in diabetic polyneuropathy. There is an elevation of the sensitivity threshold and an exaggeration of the response (hyperpathy). Also in patients who are malnourished due to a nutritional deficit that can occur even in epidemic form, in malnourished populations and who respond very satisfactorily to the administration of calcium and yeast extracts, and vitamins, especially B1.

Literature

Gopalan, C.: The 'burning feet syndrome'". Indian Med. Gaz. 81:22-26, 1946.

Vernon, S.: Nutritional melalgia, a deficiency vascular disease". J.A.M.A., 143:799-802, 1950.

JOLLIFFE SYNDROME

Synonymy

S. of nicotinic acid deficiency. Nicotinic acid deficiency encephalopathy.

Syndromography

Clínica

It is an acute cerebral syndrome characterized by progressive obscuration of the mind that progresses to coma, rigidity, extrapyramidal tremor (cogwheel sign) and uncontrollable grasping and sucking reflexes, associated with manifestations of polyneuritis and oculomotor signs of central neuritis.

Signs and symptoms of deficiencies of other vitamins in the B and vitamin C complex may additionally be observed, such as bleeding in the gums, ecchymosis and dermatitis, diarrhea and polyneuritis.

Paraclinical examinations

Blood. Low values of nicotinic acid and other B complex vitamins (niacin, B1).

Syndromogenesis and etiology

This syndrome is caused by an acute deficiency of nicotinic acid.

It is observed in Wernicke's disease, pellagra, scurvy, alcoholism and malnutrition in general.

Literature

Jollife, N., K.M. Bowman: L.A. Rosenblum and H.D. Fein: "Nicotinic acid deficiency encephalopathy." J.A.M.A., 114:307-312, 1940.

KWASHIORKOR SYNDROME

Synonymy

S. of weaning. S. of malignant malnutrition.

Syndromography

Clinic

The syndrome is observed in infants and young children (it is rarely observed in adults, only in a certain form, especially associated with alcoholism), after "weaning" when they are maintained for some time on a diet very low in protein content. It is characterized by growth retardation, apathy, irritability, crying spells and diarrhea.

At first glance the child seems well nourished because the fat deposits remain more or less normal.

Physical examination. Edema, pigmentation and peeling of the skin are observed, the hair lightens in color from black to brown and later to red, and there is marked non-painful hepatomegaly.

Paraclinical examinations

Blood. Anemia, hypoproteinemia of all fractions.

Liver biopsy. Massive fatty infiltration of the liver.

If the cause remains the same, the autopsy also reveals atrophy of the pancreas and wasting of muscle mass.

Syndromogenesis and etiology

The syndrome is caused by primary nutritional protein deficiency.

Literature

Adams, E.B.; JN. Scragg; B.T. Naido, et al.: "Observation on the etiology and treatment in Kwashiorkor." Brit. Med. J., 3:451-454, 1967.

Williams, C.D.: Kwashiorkor. J.A.M.A., 153:1280-1285, 1953.

ACANTHOSIS NIGRICANS- -ADENOCARCINOMA SYNDROME

Synonymy

S. of malignant or adult acanthosis nigricans. Acanthosis nigricans

Syndromography

Clinic

It begins in adulthood and is characterized by:

1. Cutaneous manifestations: skin alterations, characterized by papillomatosis and grayish-brownish or black hyperpigmentation in the armpits, neck, nape of the neck, genitals and other flexural areas. There may be palmoplantar hyperkeratosis.
2. Oral manifestations: hypertrophy and elongation of the papillae on the dorsum and lateral edges of the tongue. The lips, especially the upper one, may be enlarged and present filiform or papillomatous excrescences.
   The oral mucosa may have a white and velvety appearance.
3. Adenocarcinoma of the gastrointestinal tract, lung or breast.
   Less frequently from other organs: gallbladder, pancreas, uterus and ovaries. Occasionally: testicular carcinoma and lymphomas. 60% of cases correspond to gastric cancer.

Acanthosis lesions can appear before, at the beginning or during the neoplasia and increase with its development. We must also point out that when the neoplasia is eradicated, the lesions disappear.

It is good to point out that there is another type of acanthosis called: true benign acanthosis nigricans, which begins after birth, in childhood or puberty. It is not accompanied by endocrine and neoplastic lesions, and is inherited in a dominant manner with almost complete penetrance. It may remain stationary for several years or undergo complete regression.

There is another variety of acanthosis that is part of Bloom syndrome, congenital lipodystrophy and Rud syndrome.

Paraclinical examinations

Those necessary for the diagnosis of visceral carcinomas must be performed. Additionally, study of the response to insulin.

Syndromogenesis and etiology

The etiology is genetic and is transmitted by autosomal dominant inheritance; For acanthosis nigricans associated with insulin resistance, a mutation in the insulin receptor gene has been reported.

Literature

Curth, H. O.: “Die Probleme der Acanthosis Nigricans.” Hautarzt, 15:433-439, 1964.

Schwenk, W.E. et al.: "Familial insulin resistance and acanthosis nigricans: presence of a postbinding defect." Diabetes, 35:33-37, 1986.

Tasjian, D. and M. Jarrat: Familial acanthosis nigricans". Arch. Derm., 120:1351-1354, 1984.

ENTEROPATHIC ACRODERMATITIS SYNDROME

Synonymy

S. of Danbolt-Closs. S. de Brandt. S. of acrodermatitis enteropathica- zinc deficiency.

Syndromography

Clinic

It is observed in both sexes and begins in childhood between weeks and 18 months after birth, and is accompanied by gastrointestinal disorders, especially intermittent diarrhea. In the acute phase there may be psychological alterations of a schizoid type.
Growth retardation and photophobia.

Physical examination. The primary eruption is vesiculobullous, symmetrical and grouped. It is located around the natural orifices (mouth, anus, bulb) and in the eyes, occiput, elbows, knees, hands and feet. Scabs subsequently form, and the plaques can become psoriasiform lesions. Paronychia lesions are common. Alopecia is constant and hair from the eyebrows and eyelashes can also be lost, and may be accompanied by lesions characteristic of blepharitis and conjunctivitis. There is hypoplasia of the thymus.

The evolution is intermittent and during exacerbations there is mental depression, with more or less stupidity, indifference and loss of appetite. It tends to be milder with increasing age.

Paraclinical examinations

Skin biopsy and study of digestive enzymes. They are important, Candida albicans is frequently found in lesions.

Blood. Very low zinc and alkaline phosphatase concentration in serum.
Urine. Very low concentration of zinc.

Syndromogenesis and etiology

It has been considered that it is due to an autosomal recessive inheritance, since familial cases have occurred.
It is due to the absence of a low molecular weight protein that constitutes the zinc binding factor, which determines poor absorption of this element. This factor is produced by the pancreas, binds to zinc in the diet and transports it into the epithelial cells.

Alkaline phosphatase is a zinc-enzyme. Treatment with zinc cures the syndrome and normalizes phosphatase levels, in addition there is a reconstruction of the thymus.

Literature

Brandt, TIH.: "Dermatitis in children with disturbance of the general condition and the absorption of food elements." Acta Dermat. Venerol., 17:513-546, 1936.
Evans, G.W. and PE. Johnson: Zinc-binding factor in acrodermatitis enteropathica." (letter) Lancet II: 1310, 1976.
Ohlsson, A.: "Acrodermatitis enteropathica: reversibility of cerebral atrophy with zinc therapy." Pediat Act. Scand., 70:269-273, 1981.

ACRODYNIA SYNDROME

Synonymy

S. de Feer. Pink's disease. Infantile acrodynia. Erythroedema. Vegetative polyneuritic neurosis. Trophodermoneurosis. S. de Swiff. S. de Swiff-Feer. Red disease.

Syndromography

Clinic

It is observed in children between 5 months and 2 years and lasts about 4 months, especially in temperate countries. It is characterized by:

1. Profound changes in temperament: irritability, marked photophobia, increased sweating and tendency to cry most of the time.
2. Skin changes: swollen, erythematous cold areas with subsequent peeling, accompanied by pain and profuse sweating. Sometimes intense itching. Hands and feet are cold, slimy, carmine or dirty red.

   These lesions most commonly occur on the hands, feet and nose.
3. Neurological symptoms: loss of superficial sensitivity, generalized hypotonia and tremor. Generalized cutaneous paresthesia.
4. Tachycardia.
5. Stomatitis: oral mucosa red and edematous, then covered with grayish white exudate; Deep ulcerations simulating necrotic stomatitis may also be observed.
   Gingival and alveolar alterations that lead to tooth loss.
6. Hypertension, hypertonia, anorexia and insomnia may be observed.

Physical examination. Cyanosis of the fingers, toes, nose, and ulcerations and gangrene of the fingers are observed.

Ulcers of the oral mucosa with loss of vitality of the teeth. Rectal prolapse. Exanthems of palms and soles with exfoliation of large segments of skin. Skin erythema with intense itching. Muscular hypotonia that looks like pseudoparalysis, the child can hardly stand up.

Tachycardia, arterial hypertension and hypertrichosis.

In severe cases, hypertrichosis can develop on the trunk, face and the rest of the body. 50% of patients present ocular alterations: proptosis, lacrimation, photophobia, conjunctival injection, mydriasis, keratitis, sometimes papilledema and mild optic neuritis.

Paraclinical examinations

Urine. Determination of mercury. The normal figure is 0.001 mg/L. In these cases, figures of 0.1- -0.3 mg/L are found. It is usually accompanied by albuminuria and hematuria.

Syndromogenesis and etiology

Acrodynia syndrome is also known as calomel disease (when these products were used);
Currently, the most frequent cause is the ingestion or inhalation of mercury, contained in some household paints, or the use of mercurial ointments and preparations.

Normal values for mercury in urine are: 0.0001 to 0.001 mg/L. In these cases, more than O.1 is found, which can reach 0.3 mg of mercury per liter.

It is attributed to an allergic-toxic reaction to mercurials.

The etiology is unknown.

Bibliography

Feer, E.: "Eine eigenartige Neurase des vegetativen systems beim Kleinkinde." Erg. Inn. Med., 24:100-122, 1923.
Ree, R.B.: Dermatoses due to Physical and Environmental factors. Springfield, III., Charles C. Thomas, 1962.

ACROMLA SYNDROME

DUE TO PIGMENT INCONTINENCE

Synonymy

S. of Ito. S. of hypomelanosis of Ito.

Syndromography

Clinic

It has been reported predominantly in females, but cases have also been reported in males. The incidence was 1/1,000 new patients in a pediatric service.

It begins from the earliest years, but persists throughout life. It is characterized by the progressive loss of pigment without being preceded by inflammation. The alterations can take a linear or banded form, very well delimited and sometimes are varied macules (they do not have the pigmentation pattern observed in Bloch-Sulzberger syndrome), it is frequently associated with mental retardation, strabismus, alopecia and congenital dislocation of the hip.

Paraclinical examinations

Skin biopsy. Absence of inflammatory signs. Decrease in the number of melanin granules in the basal layer.

Syndromogenesis and etiology

The etiology is unknown. There is no hereditary history. Reviewing the literature, it is found that it constitutes a manifestation of an etiologically heterogeneous group of diseases whose common factor is the presence of two genetically different cell lines.

Literature

Hamada, T.; T. Saito; T. Sugai et al.: "Incontinentia pigmentiachromians (Ito)." Arch. Derm., 96:673-676, 1968.
Ito, M.: "Studies on Melanin: XI. Incontinentia pigmenti achromians: a singular case of nevus depigmentous systematicus bilateralis." Tohoku J. Exp. Med., suppl. 55:57-59, 1952.
Donnai, D. et al.: *Hypomelanosis of Ito: a manifestation of mosaicism or chimerism", J. Med. Genet., 25:809-8 18, 1988.

ACROKERATOELASTOIDOSIS SYNDROME

Synonymy

S. of collagen plaques of the hands. (See S. de Favré- -Racouchot.) S. of nodular elastoidosis.

Syndromography

Clinic

Small, yellow or pink, warty papules can be seen in the form of narrow bands. Sometimes telangiectasias occur in the skin of the hands, especially located on the palmodorsal edge that extends from the tip of the big finger to the radial edge of the index finger.

Paraclinical examinations

Skin biopsy. An elastic degeneration of the collagen fibers of the skin is observed in the affected regions.

Literature

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

AINHUM SYNDROME

Synonymy

S. of spontaneous dactylolysis. S. of Banko-Kerend (Sudan). S. of Sukha-Pakla (India).

Syndromography

Clinic

It is more common in black patients from Africa, India or Central America, especially around the Panama Canal zone. Some cases have been reported in whites.

It affects the toes, especially the thick one, and sometimes the hands and is characterized by a linear constriction that later leads to spontaneous amputation of that part.

Usually unilateral, although sometimes bilateral.

It begins with a keratotic-type thickening of the skin on the foot, later a transverse groove appears in the skin of the flexor aspect of the little toe, usually below the interphalangeal joint. This groove deepens and extends around the finger until both ends meet, so that the finger begins to be constricted as if by a tourniquet and causes spontaneous gangrenous amputation.

The course of the disease is slow, and amputation occurs between 5 and 10 years.

Paraclinical examinations

Skin biopsy. A constrictive band of connective tissue can be seen. The arterial walls also present alterations.

Radiology. Acrosteolysis of the affected finger is observed, especially in the middle and terminal phalanx.

Syndromogenesis and etiology

The word ainhum, of African origin, means strangulation or constricting lasso.

The etiology is unknown in true ainhum. Pseudo-ainhum is a term used in relation to certain hereditary and non-hereditary diseases in which annular constriction of the fingers occurs. This syndrome has been reported in a father and two sons, suggesting an autosomal dominant monogenic etiology.

Literature

Da Silva Lima, J.E: "Study on Ainhum annoying banda ñao described peculiarly vaça ethiopa e affecting the two minimum two fish" Gaz. Med. of Lisbon, 321, 350, 378, 410, 1867.

Da Silva Lima, J.E.: "On ainhum." Arch. Dermat., 6: 367, 1880.

Rook, A.; D.S. Wilkinson and FJ.B. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

Simon, K.M.B.: “Ainhum, a family disease.” J.A.M.A., 76:560, 1921.

CUTANEOUS ALBINISM SYNDROME

Synonymy

S. of partial albinism. S. of stains.

Syndromography

Clinic

In cases affected by this syndrome, the eyes and periorbital regions are of normal color. There are white patches due to depigmentation on the skin and hair.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance, but consanguinity plays an important role. There is a congenital deficiency of tyrosinase, an essential enzyme for the oxidation of tyrosine to dihydroxyphenylalanine (DOPA) in the synthesis of melanin. Therefore, although melanocytes are active, melanin is not deposited in premelanosomes.

EYE ALBINISM SYNDROME

Syndromography

Clínic

The skin is normal and the symptoms are limited to the eyes.

They have no pigment in the retinal epithelium, or only isolated spots.

Syndromogenesis and etiology

Two X-linked recessive forms and one autosomal recessive form are reported. It has been proposed that the genetic defect determines abnormal decussation of the optic pathway and consequently loss of ocular pigment.

Literature

Creel, D.; FAITH. Donell and C.J. Withrop: Visual system anomalies in human ocular albinos." Science, 201:931-933, 1978.

0CULOCUTANEOUS ALBINISM SYNDROME

Synonymy

S. of complete albinism. S. of perfect albinism. S. of generalized albinism. S. of universal leukopathy.

Syndromography

Clínic

This syndrome occurs in both sexes from birth. It is characterized by an almost general lack of pigmentation, which affects the skin, hair and eyes. The skin is pink and thin and very sensitive to sun exposure, which causes erythema and burns. The hairs are fine as silk and pale yellow. The eyes attract attention because the pupils are red; the irises are pink or bluish. There is photophobia, nystagmus, rapid blinking and tearing. Sometimes deafness and muteness.

Within this general syndromic picture, two types have been described more recently, which we explain below.

ALBINISM0 0CULOCUTANEOUS SYNDROME TYPE I

Synonymy

S. of tyrosinase-negative oculocutaneous albinism. S. of yellow albinism.

Syndromography

Clinic

Frequency 1/10,000. The frequency of manifestations between first cousins is 4.5%. 70% have translucent irises. Nystagmus is always present, although it is reported
There is disagreement between brothers.

Paraclinical examinations

Skin biopsy. Melanocytes with absence of melanin.

Determination of the activity of the enzyme tyrosinase.
Lack of activity

Syndromogenesis and etiology

There is a deficiency of the enzyme tyrosinase, whose gene has 5 exons. Exons 4 and 5 are located on 11 p. The genetic defect has a pleiotropic effect on the visual pathway of these patients. To date, 23 types of type I oculocutaneous albinism have been reported due to amino acid substitution that affect normal enzyme function. In all cases, the tyrosine oxidation pathway to DOPA is affected.

Literature

Giebel, L.B.; K.M. Strunk and R.A. Spritz: Organization and nucleotide sequences of the human tyrosinase gene and a truncated tyrosinase-related segment." Genomies, 9:435-445, 1945.

Hsia, D. Y.: Inborn Errors of Metabolism. Chicago, The Year Book Medical Publishers, Inc., 1960.

TYPE II OCULOCUTANEOUS ALBINISM SYNDROME

Synonymy

S. of albinoidism. S. of tyrosinase positive oculocutaneous albinism.

Syndromography

Clínica

In the white race it is difficult to distinguish these patients from tyrosinase-negative albinism (in eucaroids). In blacks, the hair is yellow and pigmented spots develop on the skin. At birth they are absolutely white and present nystagmus, then normal skin pigmentation appears and their hair is yellow, but ocular albinism persists.

Syndromogenesis and etiology

The enzymatic activity of tyrosinase is positive.
In this syndrome there appears to be a block in eumelanin formation with continued pheomelanin formation.
The existence of a different gene from that of type I albinism is assumed.

Literature

Kugelman, T.P. and E.J. Van Scott: "Tyrosinase activity in melanocytes of human albinos", J. Invest. Derma., 37:73-76. 1961.

Walpole, I.R. and M.T. Mulcaky: "Tyrosinase positive albinism with familial 46,xy,t (2;4)(931.2; q31.22) balance translocation."
J. Med. Genet., 28:482-484, 1991.

KNUCKLE PADS SYNDROME, LEUCONYCHIA AND DEAFNESS

Synonymy

Bart-Pumphrey S.

Syndromography

Clínica

The knuckle pads (heloderma) are well-defined, plaque-like fibrous thickenings formed on the extensor surfaces of the proximal interphalangeal joints of the fingers and toes, including the thumbs. They develop at any age, begin to grow at a rate of 8-12 mm in diameter over the course of a few weeks or months and become permanent.

They are flesh-colored or brown, with a slightly hyperkeratotic epidermitis that covers and adheres to them. They are part of the skin, but move freely over the underlying parts. Palmoplantar keratoderma may exist.
White discoloration of the nail (total leukonychia) is also observed.

These pads are sometimes associated with Dupuytren's contracture and camptodactyly (permanent, irreducible flexion of one or more fingers). Some authors consider this syndrome as a variant of Vörner heloderma or palmoplantar keratoderma.

It is accompanied by hearing loss (sensorineural and conduction) due to damage to the cochlea and total leukonychia.

Paraclinical examinations

Skin biopsy. According to histology, the nodules correspond to skin fibromas.

Audiogram. Hearing loss or perceptual deafness and transmission.

Syndromogenesis and etiology

Joint lesions are due to fibrinoid proliferation.
The decrease in hearing is caused by nerve defects in the cochlea or alterations in the mesodermal tissue of the middle ear. Leukonychia is caused by incomplete keratinization of the nail.

This syndrome is inherited in an autosomal dominant manner.

In the cases studied, the alterations described were observed through six generations and with transmission from male to male. The basic defect due to this mutation remains unknown. The etiology is unknown.

Bibliography

Bart, R.S. and R.E. Pumphrey: “Knuckle Pads, Leukonichya and Deafmess: Dominantly Inherited Syndrome." New. Eng. J. Med., 276: 202-207 (Jan 26), 1967.

Schwann, J.: “Keratosis palmaris et plantaris cum surditate congenita et leuconychia totali unguium.” Dermatologica, 126:335-353, 1963.

DEANCELL SYNDROME

Synonymy

Brooke-Fordyce S. S. of trichoepitheliomas. S. of cholindromas or tumors in Ancell-Spiegler turbans. S. benign hereditary epithelioma of multiple cysts. Brooke's adenoid cystic epithelioma.

Syndromography

Clinic

It predominates in the female sex and begins in late childhood or early adolescence. It is characterized by the presence of lesions in the skull (turban-forming tumors) and/or facial skin, especially in the nasolabial area and in the hairs in that area (tricoepitheliomas). This is observed in members of a family and is not accompanied by systemic manifestations.

It appears in the form of moderately hard nodules, of variable dimensions, from a grain of millet to a lentil, the color of the skin or something pale, they can be isolated or larger with a seat in the subdermal tissue if they are raised.

Paraclinical examinations

Skin biopsy. Requires the diagnosis. In the skull: islands of cells surrounded by hyaline membrane are observed with the appearance of typical cylinders. On the face: cystic-looking spaces and horn cells (tricoepitheliomas).

All lesions are benign. Skin tumors show differentiation in the direction of hairy structures, hence their name trichoepitheliomas.

Blood. Hypercholesterolemia.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. Isolated cases have been reported. In a woman heterozygous for GóPD deficiency, G6PD-deficient and non-G6PD-deficient cells were found in the same tumor, indicating a multicellular tumor origin. Although Brooke-Fordyce and Ancell-Spiegler syndromes have been reported as different entities, it is disputed that these syndromes genetically correspond to the same entity.

Literature

Ancell, H.: “History of a remarkable case of tumors, developed on the head and face. ” Med. Chir. Trans., 25:227-246, 1842.

Brooke, HG.: “Epithelioma adenoides cysticum.” Brit. J. Derm., 4:269- -286. 1892.

Fordyce, JA.: “Multiple benign cystic epitheliomas of the skin.” J. Cut. Dis., 10:459-472, 1892.

Rassmusen, J.E.: "A syndrome of trichoepitheliomas, milia and cylindromas." Arch. Derm. (Chicago) II, 610-614, 1975.

Spiegler, E.: Uber Endothelioma der Haut". Arch. Derm Syph., 50:163, 1899.

Welch, J.P.; R.S. Wells and C.B. Kerr: “Ancell-Spiegler cylin dromas (turban tumors) and Brooke-Fordyce trichoepitheliomas: evidence of a single genetic entity.” J. Med. Genet., 5:29-35, 1968.

CUTANEOUS ANERGY SYNDROME WITHOUT SYSTEMIC MANIFESTATIONS

Syndromography

Clinic

These patients have skin infections caused by fungi, which are recurrent.

Paraclinical examinations

Immunological tests at the skin level. Absence of delayed sensitivity in the skin response. Normality of systemic cellular immunity indices such as lymphocyte counts when stimulated with phytohemoagglutinins.

Syndromogenesis and etiology

In this syndrome, infections are explained by the absence of delayed sensitivity in skin tests.

Bibliography

Chilgren, R.A.; P.G. Who; H.J. Meuwissen et al.: "Chronic mucocutaneous candidiasis, deficiency of delayed hyper- sensitivity and selective local antibody defect." Lancet, 2:688-693, 1967.

Marmor, M.F. andEV. Barnett: "Cutaneous anergy without systemic disease." Amer. J. Med., 44:979-989, 1968.

ARNDT-GOTTRON SYNDROME

Synonymy

S. of scleromyxedema. S. of lichen myxedematosis. S. of papular mucinosis.

Syndromography

Clinic

It occurs in the third to fifth decades and is characterized by diffuse thinning of the skin, beneath which a lichenoid papular eruption of uniform size develops that adopts a linear distribution. These papules spread almost all over the body. These lesions can cause a distortion of facial appearance: mimic rigidity due to skin thickening and limitation of mouth opening. There may be limitation of finger movements. The skin takes on an “orange peel” appearance in some places with open pores. In addition, the skin is dry (hypohidrosis) and hypotrichosis exists.

Paraclinical examinations

Skin biopsy. Proliferation of fibroblasts and acidic mucopolysaccharide deposits in the upper part of the dermis.

Blood. Hypergammaglobulinemia, paraproteinemia.

Syndromogenesis and etiology

The primary factor appears to be paraproteinemia probably associated with individual skin reactivity. The etiology is unknown.

Literature

Gottron, H.A.: “Skleromyxodem (cine eigenartige Erscheinungsform von Myxothesaurodermie)”. Arch. Derm. Syph., 199:71-91, 1954.

Rok, A.: D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

MILIAN WHITE ATROPHY SYNDROME Synonymy

S. of white atrophy. S. of progressive fibrosis with telangiectasias.

Syndromography

Clinic

This syndrome is more common in women and is characterized by the appearance of white plaques containing punctate telangiectasias and very fine pigmented areas, which are located on the ankles.
These lesions may be preceded by poorly marked petechiae, vesicles and scabs. They are generally associated with venous incompetence (see leg stasis syndrome). In 30% of cases these lesions are followed by ulcerations.

Syndromogenesis and etiology

The cause may be a circulatory defect possibly related to primary capillaritis. The etiology is unknown.

Literature

Milian, G.: "Les atrophies cutanées syphilitiques." Bull. Soc. Fr. Derm. Syph., 36:865-881, 1929.

BAZIN SYNDROME

Synonymy

S. of erythema induratum. Indurative tuberculosis.

Syndromography

Clinic

It occurs in young women who suffer from tuberculosis. The most common location is on the calf. It is characterized by indurated, erythematous, symmetrically distributed and usually painless or slightly tender lesions, which gradually become deep nodules. The skin that covers them acquires a brownish or bluish color over the course of a few months. By palpation it is verified that they are soft in their central part; They progress until reaching ulceration.

These ulcers are irregular, deeply excavated, and have loose edges. When they heal, they leave depressed white scars that alternate with active lesions at different stages of evolution. Their number ranges from 2 and 3 to a dozen and their favorite place is the lower part of the calf.

Paraclinical examinations

Skin biopsy. Tubercles of epithelioid cells originating from the hypodermis. Sometimes there are granulomas and giant cells. There is thickening of venules and arterioles that may be occluded and appear as a cosinophilic mass surrounded by lymphocytes. In the infiltrated area, elastic fibers, collagen and fat are destroyed. Wucher atrophy (fat replacement atrophy) is followed, in turn, by fibrosis. Vascular lesions may predominate with little or no presence of granulomatous lesions.

Syndromogenesis and etiology

Until recently, Bazin syndrome was considered to be of tuberculous origin, caused by Mycobacterium tuberculosis.

Currently, some tend to consider that it is a vasculitis of the subcutaneous arteries and veins with fat necrosis.

In reality, it is considered to have an unknown etiology, and the syndrome may be due to tuberculous lesions or other causes.

Literature

Bazin, A.P.E.: Lecons théoriques et cliniques sur la scrofule, considérée en elle meme et dans ses rapports avec la syphilis, la dartre et l'arthritis. II ed., Paris, A. Delahaye, 1861, p. 668.
Montgomery, H.: Dermatopathol ogy. New York, Hocber, 1967.

BEAN SYNDROME

Synonymy

S. of bleeding blue rubber polka dots. S. of cutaneous-digestive angiomatosis. S. of nevus of the elastic blue bladders.

Syndromography

Clínic

It can be observed in both sexes during early childhood. From one to hundreds of small subcutaneous nodules of rubber-like consistency may appear, which stand out in the form of small polyps accompanied by recurrent melena and cutaneous-mucous pallor.

This syndrome is characterized by:

1. Cutaneous cavernous hemangiomas similar to rubber bladders, bluish, which are located in any part of the body, including the subungual region.
2. Sporadic night pains.
3. Intense supralesional sweating that can be very intense.
4. Hemorrhages when there are intestinal cavernous hemangiomas, which are multiple and bleed easily. They can also be located in the lung, liver, kidney, peritoneum and skeletal muscles.
5. Association with medulloblastomas and fibrosarcomas has been described.

Paraclinical examinations

Biopsy of the lesions. From the anatomopathological point of view, these are hemangiomas or venous hamaromas with a polypoid appearance of the skin, small intestine and colon, lung, kidney, peritoneum and skeletal muscles. Occasionally they are found in the liver, spleen and central nervous system.

Blood. Anemia when bleeding occurs.

Radiology. It is possible to observe lesions in affected organs.

ted.

Syndromogenesis and etiology

Cthyology is monogenic genetics and it has been proven that it is transmitted by autosomal dominant inheritance.

Literature

Bean, W.B. A. and Rathe: "Universal angiomatosis. Arch. Int. Med., 112:869-874, 1963.
Bean, W.B.: Vascular spiders and related lesions of the skin. Springfield/Il., 1985.
Walshe, M.M.; Evans, C.D. and R.P. Warin: "Blue rubber bleb naevus. Brit. Med. J., 2:931-932, 1966.

BLOCH-SULZBERGER SYNDROME

Synonymy

S. of pigmentary incontinence. S. de Bloch-Siemens. Siemens-Bloch pigmentary dermatosis S.

Syndromography

Clínica

It appears at birth within the first two years, and is 10 times more common in females than in males. It is characterized by well-defined lesions, consisting of pigmented macules spread over the trunk and upper part of the arms and legs. The pigmentation takes on whimsical shapes that resemble dendritic patterns with irregular splashes, lines, striations, spirals, polyangular spots, and spider web lesions. The drawings do not follow the lines of the nerves or the limitations of the midline. This anomaly is frequently preceded by vesicular-type inflammatory lesions, which subside and reappear in lines and plaques for weeks or months before erasing and pigmented lesions appear. The evolution is feverish. The pigmentary stage can last many years and fades afterwards without leaving any sequelae or depigmented spots. Only slight areas of atrophy marking the sites of previous lesions are sometimes observed. In this syndrome, other skin lesions may manifest, such as: pseudopeeling, atrophic alterations that resemble acrodermatitis atrophica, nail atrophy, palmoplantar hyperhidrosis, epilepsy, mental retardation, strabismus, optic atrophy, blue sclerae and exudative chorioretinitis.

Retinal vascular abnormalities and alterations of the pigment epithelium are the most important ocular lesions.

Skeletal anomalies include: syndactyly, supernumerary ribs and hemiatrophy, and shortening of the arms. There may be delayed teething, malformations or missing teeth. Corneal and lens opacity, microphthalmia, retinal pigment dystrophy and sometimes blue sclerae.

Paraclinical examinations

Blood. Eosinophilia 30-50%. Vesicles contain eosinophils: 95%.

Radiology. Dental abnormalities, microcephaly, poroencephalic guistes, skeletal abnormalities in 20%, such as vertebral anomalies, syndactyly, supernumerary ribs, etc.

Skin biopsy. In advanced periods, melanin deposits are found inside and outside the melanophores in the pigmented areas.

Karyotype. It is useful in cases of affected males.

Syndromogenesis and etiology

It is considered a monogenic disease and it has been suggested:

1. That it is inherited in an autosomal dominant manner with expression limited to the female sex.
2. That it is due to a gene linked to the X chromosome that acts as dominant in females and is lethal in males.

Consistent with this hypothesis is the fact of the report of an XXY male with the syndrome. The gene has been mapped to Xpl1. The mapping of the incontinence pigmenti gene in Xq28 has established at least two genetic forms of this entity. In a family with a grandmother and mother affected with this type II, an affected XXY male was reported.

To date, 593 females, 16 males and 44 with unspecified sex had been reported in the literature.

Literature

Bloch, B.: "Eigentumliche bisher nicht beschriebene Pigmentaffekton (Incontinentia pigmenti)". Schwiz. Med. Wschr., 56:404-405, 1926.

Cannizaro, L.A. and F. Hecht: "Gene for incontinentia pigmenti maps to band Xp11 with an (X10) (p11; q22) translocation". Clin. Gednt., 32:66-69, 1987.

Carney, R.J. Jr.: "Incontinentia pigmenti: a world satatistical analysis." Arch. Derm., 112: 535-542, 1976.

García-Dorado, J. et al.: "Incontinentia pigmenti: XXY male with a family history." Clin. Genet. 38: 128-138, 1990.

Sulzberger, M.B.: Ueber eine bisher nicht beschriebene congenitale Pigmentanonmalie (Incontinentia pigmenti)". Arch. Derm. Syph., 154: 19-32, 1928.

BUREAU-BARRIERE SYNDROME

Synonymy

S. of pseudosyringomyelia.

Syndromography

Clínic

It is seen almost exclusively in men and begins in adulthood. It is especially seen in people with chronic malnutrition and frequently alcoholics. A contributing factor is trauma to the feet. It is characterized by transient vesicular lesions that lead to single or multiple perforating ulcers on the soles of the feet. The bottom of the lesion is granular and crusted. These injuries give rise to a deformity of the feet that takes on a cubic shape. Other times it is manifested by plantar hyperkeratosis. It is accompanied by cyanosis and hyperhidrosis of the feet.

There are sensory changes characterized by loss of sensitivity to temperature; tactile and painful sensitivity are little affected (pseudosyrin-gomyelia). Preserved deep sensitivity. Tendon and plantar reflexes are normal, but Achilles areflexia and muscle atrophy are sometimes found.

Paraclinical examinations

Radiology. Arthritis in the interphalangeal and metatarsophalangeal joints with partial dislocation. In advanced stages there is bone resorption in these areas.

Cerebrospinal fluid. Normal.

Oscillometry and arteriography. They are useful.

Syndromogenesis and etiology

Trophic alterations of peripheral nerves, protein deficiencies and alcoholism are found. There are no spinal injuries.

The etiology is unknown.

Literature

Bureau, Y. and H. Barrière: "Ulcerating and mutilating trophic lesion of the lower limbs." Brit. J. Derm., 70:372-377, 1958.

BUSCHKE I SYNDROME

Synonymy

S. of adult scleroedema.

Syndromography

Clinic

It predominates in the female sex and begins at any age. There is a history of infections in the days or weeks before the clinical manifestations. It is characterized by moderate fever, malaise, myalgias and arthralgias. Shortly afterward, symmetrical indurations of the skin appear with a line separating them from healthy skin. It is located on the face and neck, and causes difficulty in opening the mouth and sometimes in swallowing. Later the injuries can affect the shoulders, arms and trunk. Occasionally there is pleuropericardial effusion, and sometimes the parotids and eyes also present alterations.

Paraclinical examinations

Erythrosedimentation. Moderately high. Cells

LE: negative.

Electrocardiogram. Transitory changes.

Skin biopsy. It shows degenerative changes in the collagen fibers, which are fenestrated. Increase in mast cells.

Syndromogenesis and etiology

It has been considered possible that it is due to obstruction of the lymphatic ducts or hormonal alterations (estrogens).

The etiology is unknown.

Literature

Buschke, A.: Ueber Scleroedem". Berl. Klin. Wschr., 39:955-957, 1902.

Piffard, H.G.: An Elementary Treatise on Diseases of the Skin, for the use of Students and Practitioners. London, Mac Millan, 1876.

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology, Philadelphia, Davis, 1968.

BUSCHKE-OLLENDORFF SYNDROME

Synonymy

S. of disseminated lenticular dermatofibrosisosteopoikilosis. S. of osteopoikilosis. S. of dermato-

Syndromography

Clinic

It presents the features of multiple cutaneous fibromas associated with osteopoikilosis. It is characterized by the presence of papular fibromas symmetrically distributed over the upper part of the torso, arms, lumbar region, buttocks and thighs. Slightly raised lesions are pea-sized, oval or almost round, and the same color as the surrounding skin; They are hard and can be felt rather than seen. These lesions can be associated with other skin lesions: stretch marks, nevi and keloids.

Accompanying these injuries is the bone lesion characteristic of osteopoikilosis, which can manifest itself as painful discomfort in the shoulders and upper arms.

Paraclinical examinations

Radiology. It shows a characteristic appearance that is manifested by numerous tiny oval condensation lesions that they follow the cje of the bone (osteopoikilosis). These lesions are observed in all bones except for: vertebral column, scapula, patella, clavicle and cananeus, and have a preference for the epiphyses.

Skin biopsy. Histology shows the typical lesion as a focus of fibromatous thickening of the cutis, without destruction of elastic fibers or inflammatory reaction.

Syndromogenesis and etiology

Buschke-Ollendorff syndrome is transmitted by autosomal dominant inheritance and cases within family members are common. There have been reports of adults affected by skin and bone lesions whose children had skin lesions from birth. The basic defect of this mutant gene is unknown.

Literature

Curth, H.O.: “Dermatofibrosis lenticularis disseminata and osteopoikilosis.” Arch. Derm. & Syph., 30:552-560 (Oct.), 1934.

CANDIDA ALBICANS SYNDROME

Syndromography

Clínic

It is characterized by crusted skin lesions that are arranged symmetrically around the mouth, nose, neck, ears, perineum and other areas.

Physical examination. It demonstrates paronychia with edema and redness of the base of the nails, hands and/or feet. This results in nail growth arrest and deformity. Loss of hair color and graying with or without alopecia are also observed. There are no diarrhea or other general symptoms.

Paraclinical examinations

Crop. Examination of the lesions demonstrates the existence of Candida albicans.

Syndromogenesis and etiology

There are other types of lesions caused by this fungus that take localized or generalized forms and can even spread throughout the body and cause serious clinical forms. The etiology is unknown.

Literature

Baird, K.H.: Unusual syndrome associated with Candida albicans infection". Pediatrics, 4:730-734, 1949.

Schultz, F.W.: Two cases of thrush with unusual symptoms and skin manifestations", Am. J. Dis. Child., 29:283. 1925.

Schultz, EW.: "Systemic thrush in childhood." J.A.M.A., 105, 650-653, 1935.

CANNON SYNDROME

Synonymy

S. of fluffy white nevus. S. of white fold gingivostomatitis.

Syndromography

Clínic

Cannon syndrome is observed in both sexes and begins at any age from childhood. It is generally asymptomatic.

It is characterized by the presence of spongy lesions in the form of thick white folds that are located in the oral and labial mucosa, occasionally in the mucosa of the anal and vaginal region.

Paraclinical examinations

Skin biopsy. Demonstrates acanthosis, parakeratosis, hyperkeratosis, and vacuolated cells.

Syndromogenesis and etiology

This syndrome is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Cannon, A.B.: "White sponge nevus of mucosa (naevus spongiosus albus mucosae)". Arch. Derm. Syph., 31:365-370, 1935.

Witkop, CJ.: C.H. Shankle: J.B. Graham et al.: "Hereditary benign intraepithelial dyskeratosis II. Oral manifestations and hereditary transmission." Arch. Path., 70:696-711, 1960.

CARDIOFACIOCUTANEOUS SYNDROME

Syndromography

Clinic

This syndrome is characterized by congenital heart defects, characteristic facies, ectodermal abnormalities and short stature.

The most common heart diseases are: pulmonary stenosis and atrial septal defect. The facies is characterized by bitemporal narrowing, hypoplasia of the supraorbital rim, antimongoloid deviation of the palpebral fissures, depression of the nasal bridge, and rotated ears with a prominent helix. The hair is sparse and fragile, the skin alterations vary from hyperkeratotic areas to a generalized alteration similar to ichthyosis.

Paraclinical examinations

Echocardiogram. Malformations are confirmed mentioned cardiovascular diseases.

Skin biopsy. Hyperkeratosis and ichthyosiform alterations are demonstrated.

Syndromogenesis and etiology

There is a clinical overlap between this entity and Nonan syndrome, but they have been classified as different entities.

Although there is no defined genetic etiology, everything seems to indicate that it is a monogenic effect due to de novo mutations in relation to advanced paternal age.

Literature

Bottani, A.: I. Hammerer and A. Schmzel: The cardio-facio-cutaneus syndrome: report of a patient and review of the literature". Europ. J. Pediat., 150:486-488, 1991.

Neri, G.M.: R. Zollino and J.F. Reynolds: "The Noonan-CFC controversy (Editorial) Am. J. Med. Genet., 39:367-370, 1991

DERMATOGENOUS CATARACT SYNDROME

Synonymy

S. de Andogsky. Neurodermatitis. S. of eczema plaques. S. of pruritic lichenoid dermatitis.

Syndromography

Clinic

This syndrome is characterized by chronic eczematous skin lesions since childhood, which develop bilateral cataracts upon reaching the third decade of life.

Skin lesions produce lichenification of the skin of the neck and flexor surfaces of the limbs, especially the elbows and knees.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Andogsky, N.: "Cataract dermatogenes." Klin. Msbl. Auge-nheilk., 52:824-831, 1914.

Thannhauser, S.J.: "Werner's syndrome (progeria of adult) and Tothmund's syndrome: two types of closely related heredofamilial atrophic dermatosis with juvenile cataract and endocrine features: a critical study with five new cases". Ann. Int. Med., 23:559-626, 1945.

CINDERELLA SYNDROME

Synonymy

S. of ashy dermatosis. S. of chronic erythema. S. figuratum. Melanodermic S. S. of ash dermatosis. S. of persistent dyschromic erytheme.

Syndromography

Clinic

It begins at any age and in both sexes. The lesions can appear on the face, trunk and limbs, in the form of pigmented plaques of ash gray color with some violet tinge and with a thin infiltrated erythematous border.

Some adopt orbicular or banded shapes, but always well defined. They grow eccentrically and adopt peculiar polyclinic patterns. Sometimes there is hypopigmentation in the plaques. No systemic manifestations occur in this syndrome.

Paraclinical examinations

Skin biopsy. It shows mild follicular hyperkeratosis and hydropic degeneration of the basal cell layer. In the upper layer of dermis there is a perivascular infiltrate of lymphocytes, histiocytes and macrophages with melanin granules. In the inactive phase of the injury. It reminds us of incontinence pigmenti syndrome.

Serological examination. To exclude the pint.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Knox, J.M.; B.G. Dodge and R.G. Freeman: "Erythema dyschromi cumperstans." Arch. Derm. (Chicago), 97:262-272, 1968.

Ramírez, C.0.: The Ashes: clinical problem. Proc. I Cent. Amer. Cong. Derm., San Salvadore, Dec. 5-8, pp. 122-130, 1952.

CRET SYNDROME

Synonymy

S. calcinosis-Raynaud-sclerodactyly-telangiectasias (CRET). In English it is known as CRST (S=sclerodactyly).

Syndromography

Clinic

More common in women and begins around age 45. It has been reported only in Caucasians. The first symptoms are, generally, hemorrhages (epistaxis, melena) and are due to the presence of: mucosal telangiectasias that are also found in the hands and feet, and suggest a hereditary Rendu-Osler-Weber telangiectasia; Raynaud's phenomenon that is usually accentuated and sometimes gives rise to ischemic necrotic ulcerations; cutaneous calcinosis in the dermal and subcutaneous layers and frequently appears as large, multiple nodular deposits over the large joints (elbows and knees). Sclerodactyly is typically associated with sclerodermatous disorders and acrosclerosis, often with dysphagia.

Paraclinical examinations

Radiology. Demonstrates subcutaneous calcinosis. Esophagus: alterations compatible with scleroderma.

Blood and urine tests. They are normal.

Skin biopsy. Similar to scleroderma, but not exact.

Syndromogenesis and etiology

It tends to be considered among collagen diseases. Some authors consider it a benign variant of scleroderma.

The etiology is unknown.

Literature

Dellipiani, A.W. and M. George: "Syndrome of sclerodactyly, calcinosis, Raynaud's phenomenon and telangiectasia." Brit. Med. J., 4:334-335, 1967.

LANUCA RHOMBODAL CUTIS SYNDROME

Synonymy

S. of Jadassohn's complexion. Cutis rhomboidalis nuchae. S. Labrador collar.

Syndromography

Clínica

The skin on the dorsal part of the neck thickens and becomes hard and leathery, exaggerating normal skin marks and forming diagonal ones. All of this, together with the tanning produced by the sun, gives a characteristic appearance that resembles geometric figures.

Syndromogenesis and etiology

This syndrome is considered due to irritation from the sun.

It is common in farmers, sailors and athletes exposed to a considerable amount of sunlight. It is part of the elastosis syndromes in which the elastic tissue of the skin is affected.

Literature

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

CUTIS VERTICIS GYRATA SYNDROME

Synonymy

S. Audry. S. of Friedrich-Erb-Arnold. S. of bulldog skin.

Syndromography

Clínica

It is observed only in men and at all ages. It is characterized by the formation of folds and furrows in the scalp, especially at the vertex of the head, but they can appear in other places such as the face. Generally in number of 2-20 and have an approximate width of 1 cm. This gives a corrugated or twisted appearance to the skull. The hair grows normally and is black.

These signs can appear in isolation in a patient, which constitutes the syndrome we study, or be part of other syndromes such as: Touraine-Sólente-Golé, Marie, idiocymicrocephaly, myxedema, melanoctic nevus. More rarely it can be associated with psychological disorders, such as schizophrenia, idiocy and imbecility.

Syndromogenesis and etiology

Some authors consider this syndrome as a reversion to a more primitive stage of animal life. The folds are similar to the folds seen in the skin of tigers, lions and bulldogs. For others, traumatic factors, inflammations, viruses, tumors, nevi and proliferative diseases are involved. It has even been suggested that there is pituitary dysfunction or thyroid aplasia. It is rarely familiar.

Two forms of cutis verticis gyrata are referred to in McKusick's catalog: one is autosomal recessive and the other is sex-linked recessive.

The etiology is unknown.

Literature

Audry, Ch.: “Pachidermie occipitale vorticellée (Cutis verticis gyrata)". Ann. Derm., 10:257-258, 1909.

Mac Gillivray, R.C.: “Cutis verticis gyrata.” Cutis, 4:1211-1215, 1968.

Polan, S. and T. Butterworth: "Cutis verticis gyrata, a review with report of seven new cases." Amer. J. Ment. Defic., 57:613-631, 1953.

Robert, A.: Journal de Chirurgie par Malgaigne, Pari, I;125-126, 1843.

DARIER-ROUSSY SYNDROME

Synonymy

S. of subcutaneous sarcoidosis.

Syndromography

Clinic

It is characterized by nodules 1-3 cm thick that appear on the trunk and limbs (rarely on the face).

They generally adopt a symmetrical arrangement and are fixed to the skin that covers them; This has a pink-violet color, but does not show a tendency to ulcerate.

Paraclinical examinations

Skin biopsy. Demonstrates granulomatous lesions with the characteristics of sarcoidosis.

Syndromogenesis and etiology

Its etiology is sarcoidosis, but it can also be caused by other causes. (See Besnier-Boeck-Schaumann syndrome.)

Literature

Darier, J. and G. Roussy: “Des sarcoides sous-cutanees; contribution á l'etude des tuberculides ou tuberculose atténuees de l'hypoderme". Arch. Med. Expér. et d'Anat., 18:1-50, 1906.

DARIER-WHITE SYNDROME

Synonymy

S. of White. Dyskeratosis follicularis vegetans. S. of keratosis

Syndromography

Clinic

It is observed in both sexes, more common in males and begins in childhood.

The most striking thing is the appearance of hyperpigmented or hypopigmented macules with tan-gray mottling or reticulated plaques that in some regions take on the appearance of a fine reticulum. There is also atrophy and telangiectasias, thus completing the picture of atrophic vascular poikiloderma. These lesions are located in the upper part of the torso, neck and face, less frequently in the limbs. The nails are dystrophic, with thinning, sharpening and distortion as a result of atrophy. Leukoplakia is mainly seen in the oral mucosa but extensive involvement and warty thickening may occur. It can take the anus and the urethral meatus.

Other manifestations are: palmoplantar hyperhidrosis, bullous conjunctivitis, gingival alterations, dysphagia due to esophageal stenosis and diverticulosis, skeletal anomalies, aplastic anemia, hypersplenism and mental retardation.

Malignant neoplasms of the skin, mouth, rectum and cervix are common.

Paraclinical examinations

Skin biopsy. At the beginning, fissures are observed that take the basal layer and later extend to the Malpighian layer. There are small groups of cells isolated from the rest, with a dark nucleus and a light cytoplasm, with characteristics of partial keratosis. There may be hyperkeratosis, parakeratosis, and acanthosis to varying degrees.

Syndromogenesis and etiology

This syndrome is transmitted by autosomal dominant inheritance. Some authors think that it is linked to sex.

Benign pemphigus, acrokeratosis verruciformis, and Darier-White syndrome have been considered to be the same entity. The basic defect of the gene is not specified.

The etiology is unknown.

Bibliography

Darier, J.: “Psorospermose folliculaire vegetante.” Ann. Derm. Syph., 10:597-612, 1889.

White, J.C.: “A case of keratosis (ichthyosis) follicularis.” J. Cutan. Genitourin. Dis., 7:201-209, 1889.

Witkop, C.J.Jr. and R.J. Gorlin: "Four hereditary mucosal syndromes. " Arch. Derm., 84:762-771, 1961.

DEGOS SYNDROME

Synonymy

S. of papulosis malignant atrophy. S. Degos-Delort-Tricot. S. fatal skin/intestinal. S. of atrophic papulosquamous dermatitis. S. disseminated cutaneousintestinal thromboangiitis.

Syndromography

Clínica

It predominates in men between 20 and 40 years old. Its first stage can take weeks or years and affects the trunk, neck and proximal regions of the limbs. It is characterized by the appearance of rounded, pale pink, edematous papules. In a few days they become umbilicates. The central depression enlarges; The center takes on a porcelain white color, which may be covered by a crust. The peripheral area becomes livid red or telangiectatic, which evolves towards atrophy.

The rash progresses in groups, in which a few new lesions appear.

Acute abdominal symptoms consist of epigastric pain, fever, and vomiting of blood. Death is due to fulminant peritonitis caused by multiple perforations of the intestine. There may be eye lesions in the form of episcleritis.

Patients survive an average of two years after the onset of the disease.

Paraclinical examinations

Skin biopsy. Vascular alterations that affect arterioles and venules with intimal proliferation, fibrinoid degeneration and thrombosis.

Syndromogenesis and etiology

It is possible that it is a clinical variety of polyarteritis, or immunological or autoimmune processes.

The etiology is unknown.

Literature

Kohlmeier, W.: "Multiple Hautnekrosen bie Thromboangiitis obliterans." Arch. Derm. Syph., 181:783-792, 1941.

Degos, R.; J. Delort and R. Tricot: Dermatitiie papulosquamense atrophiante". Bull. Soc. Franc. Derm. Syph., 49:148-150, 1942.

May, R.E.: "Degos' syndrome." Brit. Med. J., 1:161-162, 1968.

DERCUM SYNDROME

Synonymy

S. of painful adiposis. Dercum disease.

Syndromography

It predominates in women aged 40-60, that is, after menopause. It is characterized by pain in certain parts of the body

where there is fat accumulation. There is asthenia, headache, decreased sweating. Generally, patients progress towards depression and mental deterioration.

Physical examination. Accumulation of subcutaneous fat; The skin is reddish or bluish in color. Palpation is painful but there is decreased sensitivity to touch.

Syndromogenesis and etiology

It is debated whether it is an autonomous entity or part of obesity itself. It has been reported as an autosomal dominant inheritance.

The etiology is unknown.

Literature

Cantú, J. et al.: “Autosomal dominant inheritance in adiposis dolorosa (Dercum's disease)". Humangenetik, 18:89-91, 1973.

Dercum, FX.: Three cases of an hitherto unclassified affection resembling in its grosser aspects obesity, but associated with special nervous symptoms, adiposis painful". Amer. J. Med. Sci., 104:521-535, 1892.

Joseph, H.L.: “Painful adiposity (Dercum's disease).” Arch. Derm., 74:332, 1956.

SEBORRHEIC DERMATITIS SYNDROME

Synonymy

S. of Unna.

Syndromography

Clinic

It is manifested by itching that frequently affects the skull, external ear and retroauricular region and less frequently extends and is located in the back, sternal region and other areas.

Zones.

Physical examination. Scales are observed on the skull and in the areas affected by dermatitis. There may be exfoliative dermatitis with or without added secondary eczematization.

Paraclinical examinations

Skin biopsy. Hyperkeratosis and parakeratosis, intracellular and extracellular edema. Perifoliculitis and infiltration with polymorphonuclear cells, lymphocytes and plasma cells are common.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Montgomery, H.: Dermatopathology, New York, Hoeber, 1966.

Unna, P.G.: "Das sebborhoische Ekzem." Mschr. Pract. Derm., 6:829-846, 1887.

ERPETIFORM SENIL DERMATITIS SYNDROME

Synonymy

S. pemphigoid. S. parapemphigus. S. of pemphigus of old age.

Syndromography

Clinic

It is observed in both sexes and begins after 60 years of age in 80%. It is characterized by injuries non-specific urticarial or eczematous lesions on the limbs.

In the urticarial form, vesicles form after 2-3 weeks. In the eczematous type, the vesicles appear after several months. Generalized forms are reminiscent of erythema multiforme, and the vesicles contain a clear and occasionally hemorrhagic fluid. Afterwards, the lesions are reabsorbed without leaving traces, only slight hyperpigmentation.

They rarely injure the mucous membranes.

Paraclinical examinations

Skin biopsy. Subepidermal vesicles without acantholysis are observed.

Syndromogenesis and etiology

French authors consider it to be a variant of Stevens-Johnson syndrome.

The etiology is unknown.

DERMATOMYOSITIS SYNDROME

Synonymy

S. of Wagner-Unverricht. S. of gregarine polymyositis.

Syndromography

Clínica

Gregarines are protozoa parasites of insects. The predominance in the female sex is 2: 1. It begins in childhood after the age of 10 or in adults predominantly in the fourth to sixth decades.

Symptoms vary according to the location of the lesions, whether on the skin or muscle. It is accompanied by fever, general malaise and weakness, erythematous rash with eyelid and periorbital edema. The erythema generally affects the face, upper part of the trunk and arms. The hands have pink plaques, especially in the periungual regions and on the joints. The capillaries are visible under the nail. After passing the acute phase, hyperpigmented or hypopigmented spots and telangiectatic erythema remain. Other types of skin lesions may be observed: vesicular urticaria, erythema nodosum or hypertrichosis.

Muscle weakness occurs in the shoulders and hips.

Swallowing disorders, respiratory distress, and tachycardia may be present (30% of cases).

Paraclinical examinations

Skin and muscle biopsy. It is essential.

Blood. Increase in oxalacetic transaminase and erythrocyte sedimentation rate.

Erythrosedimentation. LE cells: Negative.

Electrocardiogram and electromyogram. Important.

Investigate neoplasms. Sometimes these clinical pictures are paraneoplastic manifestations that disappear when the neoplasm is removed.

Syndromogenesis and etiology

Autoimmune mechanisms associated with neoplastic diseases may occur, especially in adults.

The etiology is unknown.

Literature

Unverricht, H.: “Dermatotomyositis acuta.” Deustche med. Wschr., 17:41-44, 1891.

Wagner, E.: Fall Einer Seltener Muskelkrank-Heit". Arch. Heilk., 4:282-283, 1863.

ACUTE FEBRILE NEUTROPHILIC DERMATOSIS SYNDROME

Synonymy

S. de Sweet.

Syndromography

Clinic

It is observed in middle-aged women. An infection almost always precedes it and sometimes a high fever persists. It is characterized by a painful plaque-shaped rash located on the limbs, face and neck. Later they transform into pustules.

Paraclinical examinations

Skin biopsy. Focal infiltration of neutrophils and lymphocytes accompanied by vasodilation and moderate edema of the vascular endothelium.

Syndromogenesis and etiology

It may be due to a hypersensitivity reaction to certain infectious agents.

The etiology is unknown.

Literature

Sweet, R.D.: "An acute febrile neutrophilic dermatosis." Brit. J. Derm., 76:349-356, 1964.

ECTODERMIC ANHYDROTIC DYSPLASIA SYNDROME

Synonymy

S. de Weech. S. of hereditary ectodermal dysplasia. St. of Christ-Siemens-Touraine. Siemens S. S. of hypohidrotic ectodermal dysplasia.

Syndromography

Clinic

It may begin in the neonatal period with fever without an appreciable cause, but it generally occurs in the second year of life. It is characterized by alterations in different regions, for example:

1. Skin and faneras. The skin is thin, dry, keratotic and hairless. Hypochromic, thin, fragile, scarce or absent hair; sometimes eyebrows and eyelashes are missing. Normal or concave nails. Few tears. The fundamental skin alterations that manifest in this syndrome can also be observed in other syndromes: Berlin, Helweg-Larsen, Wedderburn.
2. Facies. Protruding forehead, saddle nose, prominent lips, small protruding chin. Hyperpigmentation around the eyes.
3. Mouth. Dental delay with hypodontia or anodontia and conical teeth; dry mouth and nose.
4. Low size.
5. Occasional anomalies. Decreased taste and smell, upper respiratory infections and/or asthma attacks, eczema, hypoplasia of the basal turbinates; mammary and mammillary hypoplasia or aplasia. Sometimes mental retardation.

An autopsy in a patient has shown the absence of glands in the mucous membranes of the pharynx, larynx, trachea and bronchi, which explains the susceptibility to respiratory infections.

Paraclinical examinations

Skin biopsy.

Syndromogenesis and etiology

The gene responsible for this defect is transmitted following a recessive inheritance linked to the X chromosome, although several families with autosomal recessive inheritance have been reported. The gene has been located in Xql1-q21.1. Among the reported patients, only 10% are female.

The fundamental skin alterations seen in this syndrome can also be seen in Hellweg-Larsen and Wedderburn syndromes.

Literature

Perlman, H.H.: Pediatric Dermatology. Ed. Year Book Pub. Chicago, 1969. p. 394.

Richards, W. and M. Kaplan: “Anhydrotic ectodermal dysplasia. "An unusual case of pyrexia in the newborn". Am. J. Dis. Child., 117:597, 1969.

Weech, A.A.: "Hereditary ectodermal dysplasia (congenital ectodermal defect). A report of two cases." Am. J. Dis. Chil., 37:766, 1929.

Zonana, J. et al.: "X-linked hypohidrotic ectodermal dysplasia: localization whithin the region Xqll-q21.1 by linkage analysis and implications for carrier detection and prenatal diagnosis". Am. J. Human Genet., 43:75-85, 1988.

ECTODERMIC HYDROTIC DYSPLASIA SYNDROME

Synonymy

S. of Berlin. S. of Clouston.

Syndromography

Clinic

It is observed in both sexes and begins in childhood. It is characterized by nail dystrophy that can be manifested by thinning, striations, discoloration, short nails and these may be the only manifestations. Paronychia can also occur with infections. The skin around the nails, finger joints, knuckles, knees and elbows may be thick. There is palmoplantar hyperkeratosis.

Hyperpigmentation of the skin especially in the joints. In complete forms the hair is fine and spaced or absent, and generally presents defects at puberty. In men, microgenitosomia, hypospadias, testicular hypoplasia. Sweating is increased in some regions, normal in the rest;
delay in primary and secondary dentition; physical development is normal. Mental development is normal or somewhat delayed.

Paraclinical examinations

Skin biopsy. Presence of eccrine sweat glands that are active, unlike the anhidrotic form in which the glands are greatly reduced or absent, so patients cannot sweat. Hyperkeratosis.

Hair study. Optical microscopy shows disorganization of the fibrillar structure and reduced birefringence.

Syndromogenesis and etiology

The syndrome is transmitted by autosomal dominant inheritance. The basic defect corresponds to a molecular abnormality of keratin due to a gene mutation.

Literature

Berlin, Ch.: "Congenital generalized melanoleucoderma associated with hypodontia, hypotrichosis, stunted growth and mental retardation occurring in two brothers and two sisters."
Dermatologica, 123:227-243, 1961.

Bixler, D. and R.A. Patel: "Clouston syndrome: a rare autosomal dominant trait with palmoplantar hyperkeratosis and alopecia."
(Abstract) Am. J. Human. Genet., 47 (suppl.): 1990.

Clouston, H.R.: "A hereditary ectodermal dystrophy." Canada Med. Assoc. J., 21:18-31, 1929.

Gold, R.J.M. and C.R. Scriver: Properties of hair keratin in an autosomal dominant form of ectodermal dysplasia". Am. J. Hum. Genet., 24:549-561, 1972.

CONGENTTA DYSKERATOSIS SYNDROME

Synonymy

S. de Zinssers. Cole S. S. of Engman.

Syndromography

Clinic

This syndrome is almost exclusive to the male sex.

Starts between 5-13 years old. It is characterized by dysonychia with complete destruction and recurrent suppurative paronychia. Later, a grayish pigmentation develops in the shape of a reticulum, more intense on the neck and thighs, which envelops the entire trunk. In addition, areas of atrophy and telangiectasia are present. The face is red with areas covered with atrophic macules. There are macules on the posterior surface of the hands and feet, and palmoplantar keratosis with hyperhidrosis and vesicles.

At the level of the mucous membranes there are small erosive lesions with leukoplakia, especially in the oral mucosa and conjunctiva, with excessive tearing. Defects are also observed in the teeth. Areas of alopecia with scarring hypotrichia. Psychic and mental development may be delayed.

The dermatological triad: reticulated hyperpigmentation, nail dystrophy and mucosal leukoplakia, is important for the diagnosis.

Paraclinical examinations

Blood. In some cases Fanconi type anemia or pancytopenia. (See S. de Fanconi.)

Skin biopsy. The changes found are not specific. These are: parakeratosis, hyperkeratosis and acanthosis. The gastrointestinal tract and mouth show similar lesions.

Syndromogenesis and etiology

This syndrome is transmitted by sex-linked recessive inheritance, but there are cases with consanguineous parents that suggest inheritance autosomal recessive. An increase in chromatid breaks and gaps has been found after X-radiation during the G2 phase of the cell cycle.

Literature

Cole, H.N.; JE. Rauschkolb and J. Toonmey: *Dyskeratosis congenita with pigmentation, dystrophia unguis and leukokeratosis oris". Arch. Derm. Syph., 21:71-95, 1930.

De Bausche, D.M.; P.G. Shashidhar; W.S. Stanley: “Enhanced G2 chromatid radiosensitivity in congenital fibroblast dysheratosis.” Am. J. Human. Genet. , 20:85-87, 1985.

Lantigua, A.: “Consanguineous parents in a case of congenital dyskeratosis.” Not published. Dept. Genetics Inst. Sup. Cienc.
Med. Havana. Cuba. 1993.

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

Zinsser, E.: "Atrophia cutis reticularis cum pigmentatione, dystrophia unguium et leukoplakia oris (poikilodermia atrophicans vascularis Jacobi)". Ikonogr:. Derm. (Kyoto), 219-233, 1906.

DREUW SYNDROME

Synonymy

S. of alopecia parvimaculata.

Syndromography

Clinic

It is seen in both sexes and at all ages. It is characterized by the rapid development of small, irregular patches of alopecia.

Paraclinical examinations

Skin biopsy. Infiltration of lymphocytes is observed around the hair follicle, which takes the bulb0, followed by scleroatrophic changes. The sebaceous glands and hair follicles disappear but the sweat glands are intact.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Dreuw, M.: Ueber epidemische Alopecia"; Vorlaufige Mitteilung. Monatsh. F. prakt. Dermat., 51:18-22, 1910.

Hofer, W.: "Sporadisches Auftreken von Alopecia parvimaculata. " Derm. Wschr., 149:381-386, 1964.

DUHRING-BROCQ SYNDROME

Synonymy

S. de Brocq-Duhring. S. dermatitis herpetiformis.

Syndromography

Clinic

It has a predominance of 2:1 in men. The onset of this syndrome is between 20-55 years; occasionally in children over 5 years of age and may be gradual or acute. It is characterized by a pleomorphic and symmetrical eruption of the skin (erythema, urticaria, papule, vesicle or flictena), which is easily abraded; sometimes eczematous and exudative or lichenified. Lesions leave pigmented spots

(50% of cases). These lesions predominate on the extension surfaces of the limbs, elbows, knees, buttocks, frequently affecting the axillary folds, shoulders, trunk and facies, rarely the mucous membranes.

Paraclinical examinations

Skin biopsy. Accumulation of neutrophils and eosinophils within the dermal papillae. In the vicinity of the beginning blisters, there are formations in the form of subepidermal vesicles. There is moderate acantholysis.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Duhring. L.A.: Selected Monographs on Dermatology. London, New Sydenham Soc., 1893.

EHLERS-DANLOS SYNDROME

Synonymy

S. of hyperelastic skin. S. of Danlos. S. de Meekein-Ehlers- -Danlos. S. of arthrochailasis. S. of dermatorrhexis. S. of lax complexion. S. of congenital mesodermal dystrophy. S. of generalized elastic fibrodysplasia.

Syndromography

Clinic

There may be slight or incomplete forms. In childhood, growth and development are poor.

Delay in standing or walking, easy falls and tendency to fractures.

Sometimes the manifestations appear in the mature adult. There is a tendency to hemorrhage and leg cramps, especially at night.

Physical examination. Fragile and/or hyperelastic skin, which can be stretched several centimeters and returns to its place when released. Over time and depending on the clinical form of this syndrome, the skin can lose its elasticity and form folds, which on the hands and feet give the impression that the patient is wearing moccasins or gloves. The fragility of the skin frequently leads to petechiae and/or ecchymosis, sometimes hematomas that are organized into nodules and can calcify and simulate a tumor. The scars are thin and fragile, brown in color and are frequently associated with telangiectasias. Wounds heal slowly. The joints are hypermobile, and hemarthroses and dislocations are frequent. Sometimes this syndrome is associated with flat feet.

There are genu recurvatum, kyphoscoliosis and bone malformations. Different malformations of the heart, lung and gastrointestinal tract may also be present.

Syndromogenesis and etiology

Various types have been identified in this syndrome. Initially Beighton classified them into 5 types based on the main molecular defect that exists in this syndrome. They are:

Type I. Severe. It is the classic form of the disease, with joint hypermobility and characteristically velvety and hyperextensible skin. Autosomal dominant inheritance. The molecular defect is unknown.

Type II. Mild. Similar to type I, but less severe. Autosomal dominant inheritance. The molecular defect is unknown.

Type III. Benign. Joint hypermobility is more notable than skin changes. Presence of periodontitis. Autosomal dominant or recessive inheritance. The molecular defect is unknown.

Type IV. Ecchymotic or aortic. It is characterized by noticeable thinness of the skin and a predisposition to sudden death due to rupture of large blood vessels or vessels of the large intestine. Sex-linked inheritance (X chromosome). Defect in the synthesis or structure of type II collagen. This explains the tendency for vessels that are rich in collagen to break.

Type V. Linked to sex. It is similar to type II, but is transmitted by this type of chromosome.

Additionally, other types have been described, due to the presence of biochemical defects and phenotypes that do not correspond to any of the types defined by Beighton. These are:

Type VI. It was characterized in two sisters and the molecular defect is a decreased amount of hydroxylysine as a result of a lysylhydroxylase deficiency.

Type VII. Congenital multiple arthroclysis (ankylosis). With multiple dislocations and ankylosis. Autosomal recessive or dominant inheritance. There is a defect in the conversion of procollagen to collagen.

Type VII. The skin is more fragile than hyperextensible, and heals leaving pigmented atrophic scars.

Type IX. It is an Ehlers-Danlos syndrome with abnormal copper metabolism, which resembles Menke's steely hair syndrome (some variants) and cutis laxa syndrome (some variants). Sex-linked inheritance.

Literature

Danlos, H.: “A case of lax cutis with tumeurs par contusion chronique des coudes et des genoux.” Bull. French Soc. Derm. Syph., 19:70-72, 1908.

Ehler, E.: “Cutis laxa, Neigung zu Haemorrhagien in der Haut. Lockering mehrerer Artikulationen". Derm. Z., 8:173-174, 1901.

Utto, J. and E. A. Bauer: “Diseases associated with collagen abnormalities", in collagen in health and diseases, J.B. Weiss; M.I. D. Jayson (eds) New York, Churchill Livingston, 1982, p, 289.

Wheeler, C.E. Jr:: Ehlers-Danlos syndrome. In Beeson, P.B., and McDermot, W.: Cecil-Loeb Textbook of Medicine, Philadelphia, Saunders, 1967.

ELASTOSIS PERFORANS SERPIGINOSA SYNDROME

Synonymy

S. serpiginous follicular keratosis. S. de Lutz-Mischer.

Syndromography

Clínica

This syndrome predominates in men. 90% of cases begin before the age of 30, in some patients in childhood or old age, and affects all races. The lesions are located in the neck and less frequently in the upper limbs, face, lower limbs and trunk, in that order.

They are asymptomatic or slightly pruritic. It is characterized by low-lying, erythematous and keratotic papules measuring 2.5 mm with scales in the central region that extend and adopt a serpiginous or circular pattern. They frequently appear

satellite lesions. They tend to be symmetrical and can be associated with various disorders such as Van Der Hoeve syndrome and Grönblad-Strandeberg-Touraine and Rothmund syndrome.

Paraclinical examinations

Skin biopsy. Shows the most frequent changes in the injury.

Syndromogenesis and etiology

The cytology is unknown although familial incidence has been reported.

Literature

Jones, P.E. and D.C. Smith: “Porokeratosis, review and report of cases.” Arch. Derm., 56:425-436, 1947.

Lutz, W.: "Keratosis follicularis serpiginosa." Dermatologica, 106:318, 1953.

Mehregan, A.H.: "Elastosis perforans serpiginosa." Arch. Derm., 97:381-393, 1968.

ELLIS-VAN CREEVELD SYNDROME

Synonymy

S. of chondroectodermal dysplasia. S. of ectodermal chondrodysplasia. S. of mesoectodermal dysplasia. S. of tridermal chondrodysplasia.

Syndromography

Clinic

This syndrome is characterized by postaxial polydactyly, especially in the hands: nail dysplasia (hypoplasia and koilonychia) and dental alterations. Early rash and premature exfoliation. Low stature of prenatal onset with proximal shortening of the limbs (which contrasts with achondroplasia).

Fusion of the metacarpal bones, knocking knee, due to a defect of the proximal part of the tibia. Congenital heart defects are often associated, especially atrial septal defect.

Malformations of the central nervous system have been described, such as Dandy-Walker syndrome and hydrocephalus, as well as various anomalies of the urinary system.

Paraclinical examinations

Radiology. It shows almost all the bone and/or dental alterations indicated.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance. Among the Israelites of Pennsylvania it is 5 per 1,000 births. The mutant gene produces ectodermal and chondral dysplasia, which causes insufficiency of the epiphysis to produce columnar cartilage with deformity of the proximal metaphysis of the tibia, accelerated bone maturation and fusion in the hands and feet.

Literature

Ellis, R.W.B. andS. Van Creveld: "A syndrome characterized by ectodermal dysplasia, polydactyly. Chondrodysplasia, and congenital morbus cordis: report of three cases." Arch. dis. Child., 15:65-84, 1940.

McKusick, V.A.; J.A. Egelnad; R. Eldridge et al.: "Dwarfism in the Amish: the Ellis-Van Creveld syndrome." Bull. Johns Hopkins Hosp., 115:306-336, 1964.

Rosemberg, S., et al.: "Chondroectodermal dysplasia (Ellis Van Crevel) with anomalies of CNS and urinary tract." Am. J. Med. Genet., 15:291-295, 1983.

HEREDITARY LETAL EPIDERMOLYSIS BULLOSA SYNDROME

Synonymy

S. of Herlitz.

Syndromography

Clinic

Presence of bullous (flictenous) skin lesions from birth, which are observed in the navel, nails, face, scalp and limbs. Bullous lesions in the mucous membranes, especially oral (especially in the palate at its junction with the velum) due to suction. Fever.

Loss of fingernails and fingernails. It is fatal before reaching 3 months, due to dehydration and sepsis, in 94% of affected children.

Paraclinical examinations

Skin biopsy. Presence of flictenae at the dermoepidermal junction. Regeneration of basal cells in ulcerated areas. Absence of inflammatory reaction and edema.
Characteristic vacuolization of the basal portion of the germinal layer of the epidermis.

Blood. Severe and progressive hypoalbuminemia.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Herlitz, G.: "Kongenitaler nicht syphilitischer Pemphigus Eine Ubersicht nebst Beschreibung einer neuen Krankheits-Form (Epidermolysis bullosa hereditaria lethalisy'. Acta Paediatr., 17:315-371, 1935.

Roberts, M.H.; D.R.S. Howell; J.L. Bramhall and B. Reubner: Epidermolysis bullosa lethalis." Pediatrics, 2:283-290, 1960.

EPIDERMOLYSIS BULLOSA SIMPLE SYNDROME

Synonymy

S. of localized epidermolysis bullosa. S. de Fox. S. de Kobner:.

Syndromography

Clinic

This syndrome begins in childhood although it does not present symptoms except for pain and discomfort when the bullae rupture, which do not leave scars or pigmentation and heal in a few days. These flictenous elevations in the hands and feet develop at the slightest trauma. Hyperhidrosis of the hands and feet is frequently associated. Flictenae reappear especially when in contact with warm water.

Paraclinical examinations

Skin biopsy. Subepidermal bullae, containing a clear, fluid fluid. Little inflammatory reaction when

there is associated infection. The elastic tissue presents alterations but is not destroyed. It is possible to differentiate this lesion from porphyria or drug reaction or other forms of epidermolysis bullosa.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Elliot, G.T.: Two cases of epidermolysis bullosa." J. Cutan. Genitourin. Dis. 13:10-18. 1895.

Montgomery, H.: Dermatopathology. Harper-Row, New York, 1967.

CHRONIC MIGRATORY ERYTHEMA SYNDROME OF AZFELIUS

Synonymy

S. de Lipschutz. S. of erythema chronica migrans.

Syndromography

Clinic

It is observed in both sexes and at all ages. It is characterized by a single or multiple, annular eruption that grows centrifugally and expands, affecting large areas.

In some cases there are symptoms and signs of meningitis.

Syndromogenesis and etiology

It is due to the bite of ticks that cause infection by rickettsiae or spirochetes.

Literature

Afzelius, A.: "Erythema cronicum migrans." Acta Derm. Vener. (Stockholm), 2:120-125, 1921.

Hellerstom, S.: Erythema chronicum migrans Afzelius". Acta Derm. Vener. Stockh., 11:315-321, 1930.

ERYTHEMA GYRATUM REPENS SYNDROME

Syndromography

Clinic

It is characterized by lesions spread throughout the body, arched, concentric, reminiscent of the grain of wood (cypress bark).

Paraclinical examinations

The necessary tests must be performed to rule out some type of neoplasia, as it is almost always associated with them.

Syndromnogenesis and etiology

This syndrome is associated with visceral neoplasms. It is assumed to represent a hypersensitivity reaction to a component of the malignant tumor.

Literature

Gammel, J.A.: “Erythema gyratum repens, skin manifestations in patient with carcinoma of breast." Arch. Derm. Syph. , 66:494-505, 1952.

Rook, A.: D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Davis, Philadelphia, 1968.

Rothman, S.: “Uber Hauterschein-ungen bei bosartigen Geschwulsten innerer Organe.” Arch. Dermat. U. Syph. , 149:99-123, 1925.

ERYTHEMA NODOSUM SYNDROME

Synonymy

Dermatitis contusiformis S.

Syndromography

Clinic

It occurs at any age but is rare in children and elderly people, it is more common in women. It is characterized by painful nodules distributed in the anterior tibial region, on the extension surface of the forearms, on the back, arms and face. Occasionally it is accompanied by fever, general malaise and arthralgia.

Physical examination. Lesions of different sizes are observed, some of which can grow to a diameter of 2-5 cm. They are red and warm to the touch. The color can regress and take on a dark red, gray, yellowish appearance.

The lesions disappear in a few days or last up to 3 weeks, and are frequently associated with cervical lymphadenopathy.

Paraclinical examinations

It should be investigated for the primary disease.

Blood. Moderate anemia, accelerated erythrocyte sedimentation rate.

Urine. Albuminuria.

Chest radiology. Hilar lymphadenopathy is found.

Skin biopsy. Infiltration of polymorphonuclear cells in the perivascular region of the nodules. Dilation of the vessels and edema in that area.

Syndromogenesis and etiology

This syndrome can be caused by a hypersensitivity reaction secondary to numerous processes: viral, bacterial, fungal and drugs.

The etiology is unknown.

Literature

Wheeler, C.E.Jr.: "Erythemas." In Beeson, P.b., and McDermott, W., ed.: Cecil-Loeb Textbook of Medicine, ed. 12, Sounders, Philadelphia, Saunders, 1967.

PALMAR ERYTHEMA SYNDROME

Synonymy

S. of palmoplantar erythema. S. of symmetrical acroerythema.

S. de Lane. S. of hereditary palmar erythema. S. of red palms.

Syndromography

Clinic

It is evident at birth, in early childhood or in adulthood. It is characterized by symmetrical, stable and permanent erythema of the palms (located especially in the thenar, hypothenar and pulp regions of the fingers) and also on the soles (heel, plantar surface and marginal regions of the foot). There are no signs of sympathetic excitability, such as changes with temperature or emotions.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance and has been found in several generations with transmission from the father affecting several children.

The difference with acquired red palms, which are seen in liver diseases, mainly liver cirrhosis, in emphysematous or chronic lung patients and in smokers, must be specified.

The etiology is genetic.

Literature

Lane, J.E.: "Erythema palmare hereditarium (red palms)." Arch. Dermat. Syph., 20:445-448, 1929.
Montgomery, H.: Dermatopathology. New York, Hoeber, 1967. p. 66.
Olivier, J.: "Erythema palmo-plantaire hereditaire. Maladie de Lane." Arch. Belg. Derm. Syph., 12:202-207, 1956.
Poinso, R.; E. Calas; A. Stahl, et al.: "Le syndrome des paumes rouges." Presse Med., 61:275-278, 1953.

ERYTHRODERMA SYNDROME

Synonymy

S. of exfoliative dematitis.

Syndromography

Clinic

This syndrome is characterized by a generalized scaly erythematous rash that affects the entire surface of the skin, and has important implications in General Medicine due to the systemic effects caused by massive and continuous exfoliation of the skin. The intensity of the metabolic response to exfoliation depends, to a large extent, on the duration and severity of the process. Serious metabolic effects occur in chronic exfoliative dermatitis, when the peeling rate reaches 17 g/m² in 24 hours. In these cases, patients present a negative nitrogen balance, edema, hypoalbuminemia, and muscle wasting. Also important in them is the great extrarenal loss of water due to the defective skin barrier, which allows a considerably greater transepidermal loss of water.

About 60% of patients with exfoliative dermatitis heal in 8 to 10 months, 30% die, and 10% have a persistent problem that does not respond to treatment.

Syndromogenesis and etiology

This syndrome is an important dermatological complication that can be the result of the extension of a drug reaction, or the generalized spread of a pre-existing dermatitis, such as psoriasis, atopic dermatitis, or appear together with lymphoma or leukemia, on which its prognosis also depends.

ATOPIC ERYTHRODERMA SYNDROME

Synonymy

Allergic dermorrespiratory S.

Syndromography

Clinic

It is observed in both sexes and after 2 months of age, rarely before. The antecedent of history is frequent family with atopic disorders. It is characterized by erythema, papules and vesicles on the cheeks, forehead, antecubital region and popliteal flexure accompanied by severe pruritus. This syndrome is frequently associated with rhinitis and asthma.

Syndromogenesis and etiology

It is considered that this syndrome may be due to an association of some congenital factors that predispose to these reactions, to which precipitating factors are added.

The etiology is unknown.

ICHTHIOSIFORM ERYTHRODERMA SYNDROME

Synonymy

S. of fetus in harlequin. S. of malignant keratoma. S. of congenital ichthyosis (Riecke type I and type II).

Syndromography

Clinic

We will describe the two types separately:

Type I. Fetal ichthyosis. Children at birth (sometimes premature) appear to be enclosed or encapsulated in a thick white skin. They have difficulty breathing and swallowing. (See collodion baby syndrome.) Most die within 3-4 days.

Type I. Ichthyosiform erythroderma. It occurs at birth or on the first day of life, occasionally later.

It is characterized by generalized erythema with marked thickening of the skin, with peeling and crusting; edema. Lesions may be localized; erythema located in half of the body (harlequin fetus) or take up the entire body surface and therefore have different degrees of severity that can reach total encapsulation. The hair may be normal, hypertrichosis or there may be alopecia. The nails are normal.

Paraclinical examinations

Skin biopsy. Thickening of all layers of the skin. Increase in the rate of mitosis in cells and perivascular infiltration by lymphocytes.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Lenz, C.L. and J. Altman: "Lamellar ichthyosis". Arch. Derm., 97:3-13, 1968.
Seeligmann, E.: De Epidermis, Imprimis Neonatorum Desquamatione. Inaugural, Dissertation, Berlin, 1841.

DERMAL ERYTHROPOIESIS SYNDROME

Syndromography

Clinic

This syndrome is observed in the newborn and is characterized by a hemorrhagic-purpuric rash. Each lesion is 2-7 mm in diameter with a dark blue or magenta color; These lesions return slowly and disappear in 3-4 weeks.

It is accompanied by hepatomegaly, splenomegaly, jaundice (not constant) and lymphadenopathy.

Paraclinical examinations

Blood. Anemia, reticulocytosis, thrombopenia and sometimes hyperbilirubinemia.

Skin biopsy. Aggregates of large nuclear cells, erythroblasts in different stages of maturation to anucleated erythrocytes. Absence of myeloid elements (megakaryocytes, lymphocytes, etc.).

Syndromogenesis and etiology

It is associated with intrauterine viral infections. Cytomegalovirus has been demonstrated in some cases. Rubella should be considered as a possible etiology in other cases.

Literature

Brough, A.J.; D. Jones; RH. Page, et al.: "Dermal erythropoiesis in neonatal infants." Pediatrics, 40:627-635, 1967.
Dieterich, H.: Studien uber extramedullare Blutbildung bei chirurgichen Erkankungern". Arch. Klin. Chir., 134, 166-175, 1925.

SYSTEMIC SCLEROSIS SYNDROME

Synonymy

S. of progressive systemic sclerosis. S. of systemic scleroderma. S. of acrosclerosis

Syndromography

Clínica

It is a condition that affects countless organs and is characterized by inflammatory, vascular and fibrotic changes of the skin (scleroderma), and internal organs, particularly the gastrointestinal tract, lungs, heart and kidneys.
There are two predominant forms: visceral and cutaneous, which frequently coexist during the course of the condition, but which can occur in isolation. It is not always progressive and there are cases in which after many years the skin returns to normal.

It predominates in women (4:1) and its onset is generally around the fourth decade, and later in men.

Raynaud's phenomenon is very important, as it occurs in 90% of patients and precedes the onset of the disease by several years. These patients show triphasic color changes: pallor, cyanosis, blushing, in this order. If the patient presents all or any of these changes, Raynaud's phenomenon must be considered.

Less frequently, there is swelling of the fingers and hands in the initial stages and firm, thickened, leathery-looking skin, fixed to the subcutaneous tissue in more advanced stages with limitation of joint movement. There are ulcerations on fingers and legs. Hair and hair are lost. In some patients, calcifications are found and telangiectasias appear over time.

The coexistence of calcinosis, Raynaud's phenomenon, esophageal hypomotility, sclerodactyly and telangiectasia has been called CRET syndrome, which from the beginning was considered a benign form of scleroderma.

Other symptomatic elements are: weight loss, dyspnea, gastroesophageal reflux, diarrhea or constipation and abdominal pain.
terminal.

Physical examination. Changes in the skin of the face and hands, and the lesions can extend to the arms, chest and other parts of the body. The facies is typical, smooth, with loss of wrinkles. The thickened skin, the nose decreases in size, radial grooves appear around the mouth whose opening is decreased. The skin is hardened and rigid, which prevents depression of the eyelids.

There are telangiectasias. The hands are initially swollen, then they atrophy and painful ulcers, gangrene, and retraction of the hands in semiflexion appear. The same thing happens on the feet, but the injuries are less severe. There may be localized or generalized calcinosis. There is gastroesophageal reflux, dysphagia, and cramping pain in the abdomen that are accompanied by malabsorption syndrome. In the respiratory system there are: dyspnea, dry or productive cough, cyanosis and tendency to recurrent pneumothorax. Cardiovascular symptoms: arrhythmia and cardiomegaly. Osteomioarticular: muscle weakness, arthralgias and changes typical of rheumatoid arthritis. Signs of renal failure are common in 50% of patients and generally lead to death: hypertension, hyperazotemia, proteinuria, and sediment abnormalities accompanied by microangiopathic hemolytic anemia, which is caused by deposits.
fibrin sites in renal arterioles.

Paraclinical examinations

Skin and muscle biopsy. It shows sclerotic changes in the skin, vascular intimal proliferation with perivascular hyalinization and lymphocyte infiltration. Thrombotic occlusions are observed. There is a large number of collagen fibers that form compact bundles parallel to the skin and penetrate into the subcutaneous tissue, fixing the skin to it.

Blood. Elevated erythrocyte sedimentation rate and increased globulins (50%), cold agglutinins (30%), positive rheumatoid factor (30%), antinuclear antibodies (78%).

Urine. Proteinuria.

Radiology. The bones and joints show changes similar to those of rheumatoid arthritis. Digestive tract: esophageal dilation, with stenosis and abnormal peristalsis. Stomach and duodenum less affected. Colon sometimes with an appearance similar to ulcerative colitis. Chest: diffuse reticular infiltration and often cystic changes in the lower parts, pleuritis and pneumonia frequently.

Electrocardiogram. Presents abnormalities.

Kidney and respiratory tests. They are altered.

Syndromogenesis and etiology

It is debated whether this syndrome falls into the group of collagenous or autoimmune

It is considered that the primary phenomenon in this syndrome is the injury of the endothelial cells of the vascular intima in the fine arteries and capillaries, which would trigger a succession of alterations that lead first to an increase in permeability and subsequently to their occlusion. This explains why, in its incipient phase, sclerosis is accompanied by edema of the "target" organ followed by fibrosis and later-onset ulcerated lesions and telangiectasias, as well as gangrene in some areas.

Hypergammaglobulinemia and antinuclear antibodies allow us to consider that cell-mediated immune and humoral phenomena exist.

The etiology is unknown.

FABRY SYNDROME

Synonymy

S. of angiokeratoma corporis diffusum universale. S. de Fabry-Anderson. Ruiter-Pompen cardiovasorenal syndrome. S. de Sweeley-Klionsky. S. dystopic hereditary lipidosis.

Syndromography

Clinic

There is a family history of this syndrome; it predominates in men in whom the complete syndrome occurs.
In women it is only partial. Symptoms begin in childhood or adolescence, and are manifested by paresthesia in the distal region of the limbs with burning pain in the hands and feet, fever sometimes precipitated by variations in environmental temperature or by exercise; They are frequently accompanied by attacks of nausea, vomiting, abdominal pain, dizziness, headache and generalized weakness.

There is chronic respiratory obstruction secondary to the accumulation of trihexose-ceramide in the airway epithelium. Seizures, headaches, hemiplegia, vertigo.

Physical examination. As the disease progresses, typical lesions appear: eruption of telangiectatic micronodules of different sizes, located in certain areas (periumbilical, genital, buttocks and thighs), bright red or purplish in color, non-pulsatile, which decrease in color with pressure. Lack of development of beard and body hair, edema. The eyes show dilated veins in the conjunctiva and retina, frequently coneal and lens opacity. Later, high blood pressure occurs with signs of heart and kidney failure. Progressive kidney failure. Cardiomyopathy, ischemia, mitral insufficiency. Massive lymphedema of the legs.

Paraclinical examinations

Urine. Albuminuria from the beginning and vacuolated cells containing glycolipids with the appearance of "mulberry-shaped cells." Later casts, hematuriae isosthenuria. Increased excretion of trihexose-ceramide. Upon polaroscopic examination, "Maltese cross" bodies can be observed.

Blood. In advanced stages, anemia and increased urea.

Bone marrow. Vacuolated cells with glycolipids.

Slit lamp eye examination. Shows characteristic changes.

Skin biopsy. It shows telangiectasias or intradermal dilations in the papillary region, which form lacunar spaces filled with blood surrounded by acanthotic and hyperkeratotic epidermis. Blood vessels in non-telangiectatic regions have areas infiltrated by glycolipids.

Muscle biopsy. Birefringent lipid inclusions
and lamellar inclusions in lysosomes. Study of enzymatic activity in plasma, leukocytes, tears, fibroblast or lymphoblast culture. a-galactosidase A activity.

Mnolecular characterization of the mutation.

Urine metabolic tests. Proteinuria.

Syndromogenesis and etiology

It is transmitted in a sex-linked recessive manner, with total penetrance for males and partial penetrance for heterozygous females (corneal opacities). It is a glycolipid metabolism disorder with trihexose-ceramide overload and subsequent renal death, caused by an enzymatic deficiency of trihexose-ceramide-galactosyl-hydroxylase.

In exon 7, the CaT transition was found at nucleotide 1066. This change produces the substitution of arginine to tryptophan at position 356, affecting the kinetic properties and stability of the enzyme. Six substitutions have already been reported and all affect the function of the enzyme.

The basic defect is explained by a deficiency in the activity of agalactosidase A, a lysosomal enzyme whose gene has been cloned and mapped to Xq22. The enzymatic defect leads to the accumulation of the globotriaosyl-ceramide glycolipid in the plasma and lysosomes of the vascular endothelium and smooth muscle cells.

Progressive endothelial deposition in affected men produces ischemia and infarction, and leads to clinical manifestations.

older cas.

Literature

Anderson, W.: A case of angiokeratoma'". Brit. J. Dermat., 10:113- -117, 1898.

Fabry, J.: "Ein Beitrag zur Kenntniss der Purpura haemorrhaigca nodularis (purpura papulosa haemorrhagica Hebrae)". Arch. Dermat., 10:113-117, 1898.

Goodman R.M. and R.J. Gorlin: Malformations in infants and children. Illustrated guide. Madrid, Salvat, 1986, pp 160-161.

Kint, J.A.: "Fabry's disease: agalactosidase deficiency." Science, 167:1268-1 269, 1970.

Sakuraba, H. et al.: "Identification of point mutations in the alphagalactosidase. A gene in classical and atypical hemizygotes with Fabry disease." Am. J. Human Genet., 47:784-789, 1990.

Wallace, R.D. and W.J. Cooper: “Angiokeratoma corporis diffusum universale (Fabry).” Amer. J. Med., 39:656-66 1, 1965.

FAVRE SYNDROME

Synonymy

S. of stasis purple. S. of acroangiodermatitis. S. de Favre-Chaix. S. of ocher dermite.

Syndromography

Clínica

It predominates in the male sex. It is characterized by small coalescent macules that form plaques along the veins, especially the lower limbs, and extend towards the feet and upwards, their hue varies from yellow to ocher.

Syndromogenesis and etiology

It is due to stasis due to venous insufficiency. The skin in general is normal or presents discrete eczematoid changes.

Literature

Favre, M.: Nouvelle Pratique Dermatologie. V. 5, Paris, Masson, 1936, p. 113.

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology, Davis, Philadelphia, 1968.

FAVRE-RACOUCHOT SYNDROME

Synonymy

S. of solar elastosis.

Syndromography

Clinic

It is observed in people exposed to the sun for years. It generally begins between the fourth and fifth decades of life, and is characterized by yellow patches of skin with comedones, follicular cysts, and large folds of yellowish wrinkled skin that are located especially around the orbit, sometimes also on the neck and behind the ears. Elastoidosis can be accompanied by other solar degenerative manifestations such as:

rhomboidal nuchal cutis, actinic keratosis and skin carcinoma.

Syndromogenesis and etiology

The fundamental factors are the sun and perhaps the wind and the cold, especially for people who spend a lot of time outdoors in contact with these elements.

Literature

Favre, M.: "Sur une affection kystique des appareils pilo-sebaces localisee a certaines regions de la face." Bull. French Soc. Dermat. et Syph., 39:93-96, 1932.

Ritchie, E.B. and H.N. Williams: "Degenerative collagenous plaques of the hands. " Arch. Derm., 93:202-203, 1966.

IDIOPATHIC GINGIVAL FIBROMATOSIS-HYPERTHRICHOSIS SYNDROME

Syndromography

Clinic

It is observed in both sexes between birth and 9 years. It is characterized by a progressive growth of the gums (especially in the anterior part of the upper arch) that can cover the teeth completely, it can even extend the lips, but without closing them. This may be the only manifestation. On other occasions it is accompanied by marked and progressive hypertrichosis since childhood. Although they are not constant, there may be other alterations such as: mental retardation, cranial deformities, gynecomastia.

Paraclinical examinations

Gum biopsy. Hyperplasia with thin bands of collagen tissue with few fibroblasts and slight or no inflammatory reaction.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance and, although the etiology is monogenic, it has been reported that anticonvulsant medications (dilantin) can produce a clinical picture similar.

Literature

Foret, J.; P. Dodinaval; C. Forel-Kestlicher: "Hyperplasie fibreuse idiopáthique des gencives. Etude d'une famille." Genet. Hum., 13:337-350, 1964.

Vogel, R.J.: "Gingival hyperplasia and folic acid deficiency from anticonvulsive drug therapy: a theoretical relations hip." J. Theor. Biol., 67:269-278, 1977.

RADIATION FIBROMATOSIS SYNDROME

Synonymy

S. of post-radiation pseudosarcoma.

Syndromography

Clinic

It can occur between 16 months and 27 years after radiation, but usually begins after 5 years.

It is characterized by the development of a small nodule at the site of radiation.

Paraclinical examinations

Skin biopsy. Histological findings can vary markedly. In the lesions, poorly differentiated fibroblasts, interconnected connective tissue cells, etc. can be observed.

Syndromogenesis and etiology

This syndrome is considered an exaggerated response to radiation.

Literature

Pettit, V.D.; J.T. Chamness and L.V. Ackerman: "Fibromatosis and fibrosarcoma following irradiation therapy." Cancer, 7:149-158, 1954.

Samitz, M.H.: Pseudosarcoma, pseudomalignant neoplasm as a consequence of radiodermatitis." Arch. Derm., 96:283-285, 1967.

FISHER-VOLAVSEK SYNDROME

Syndromography

Clinic

It occurs in both sexes, from birth, and is associated with congenital malformations. Development is late and there are symptoms of syringomyelic syndrome.

Physical examination. Onychogryposis: poor and sparse hair, as well as eyelashes. Thinning of the terminal portion of the fingers.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Fisher, H.: Familiar hereditaries Vorkommen von Keratoma Palmare et Plantare, Nagelveranderungen, Haaranomalien und Verdickuung der Endglieder der Finger und Zehen in 5 Generationen". Dermat. Ztschr., 32: 114-142, 1921.

Volavsek, W.: "Zur der Nagelveranderungen und Palmarkeratozen bei Syringomyelie." Arch. fur. Dermat. u. Syph., 182:52-57, 1941.

FOLLICULITIS DECALVANS SYNDROME

Synonymy

S. of Auinquad. S. de Taenzer. S. sycosiform ulerythema of Unna. S. of Arnozan. S. glabrous folliculitis of the skin. S. de Feldman. S. of Little.

Syndromography

Clinic

The form that affects only the skull is seen in both sexes.

The other clinical forms are more common in men. It can generally begin in childhood and extend over the next few years. In women it begins between the third and sixth decades. Any hairy region can be affected and there are oval or rounded patches where the hair has disappeared, surrounded by perifollicular pustules. Occasionally there is pruritus. The progression of lesions is slow and takes years. This syndrome is frequently associated with atopic manifestations.

Paraclinical examinations

Crop. Demonstrates associated infections.

Skin biopsy. Follicular abscesses are observed followed by granulomatous formations with a predominance of lymphocytes, plasma cells and sometimes giant cells. Atrophic bedsores with disappearance of hair follicles and sweat glands are also detected.

Syndromogenesis and etiology

This syndrome is frequently associated with dermatitis.

seborrheic

The etiology is unknown.

Literature

Mertens, R.L.J.: «Ulerythema ophryogenes and atopy". Arch. Derm., 97:662-663, 1968.

Taenzer, P.: Uber das Ulerythema ophryogenes eine noch nicht beschriegene Hautkrankheit". Mschr. prak. Derm., 8:197-208, 1889.

FOX-FORDYCE SYNDROME

Synonymy

Fox-Fordyce disease

Syndromography

Clinic

It is rare and is not common in women during adolescence. It can be seen in men in the ratio of 10:1 to women. It is very rare before puberty or after menopause.

It is characterized by small, chronic, flesh-colored or grayish follicular papules, intensely itchy and distributed in certain regions (armpits, breasts, navel, pubis, labia majora and perineum). In these regions the hair is generally scarce. Broken hair shafts may protrude from some of the nodules, while in the center of others there are empty follicular openings that, when pressed, fill with a cloudy secretion.

Paraclinical examinations

Skin biopsy. Histology shows an obstruction of the apocrine duct at its entrance into the follicular wall. The tubular glands of the dermis are dilated.

Syndromogenesis and etiology

The etiology is related to alterations in the endocrine balance.

GIANOTTI-CROSTI SYNDROME

Synonymy

S. of infantile papular acrodermatitis. S. of infantile lichenoid dermatitis.

Syndromography

Clinic

This syndrome appears in the form of outbreaks in children from 6 months to 15 years and predominates in females.

It is characterized by the sudden appearance of flat or domed papules, round in shape, not confluent or pruritic, located on the face, neck, buttocks, limbs, palms of the hands and soles of the feet, but always sparing the trunk. The lesions last 20-40 days and disappear without peeling. The rash is accompanied by lymphadenopathy, especially in the cervical nodes, and general malaise.

Its association with viral hepatitis is common and usually persists for up to 2 months. Due to its duration, it is similar to Gibert's pityriasis rosea. Fever, rhinopharyngitis, bronchitis and gastrointestinal disorders are also common.

It is accompanied in 95% of cases by a picture of anicteric hepatitis.

Paraclinical examinations

Skin biopsy. There is a dermal infiltrate of lymphocytes, monocytes and histiocytes, with inflammatory disorders in the blood vessels of the deep dermis.

Blood. Hypertransaminasemia.

Syndromogenesis and etiology

The etiology is unknown. A viral etiology has been suggested but has not been demonstrated.

Literature

Gianotti, F: “Gianotti-Crosti syndrome.” Brit. J. Dernm., 80:342, 1968.

Zeleer, I.: "Gianotti-Crosti syndrome." Rev. Ass. Med. Argent., 73:344, 1959.

GOLTZ SYNDROME

Synonymy

S. of focal dermal hypoplasia.

Syndromography

Clinic

It is a disseminated dysplasia of the ectodermal and mesodermal structures, especially the skin and bone system. It is characterized by the presence of dark red, atrophic and sometimes cribriform plaques, almost always located on the buttocks, armpits and thighs. The lesions have a linear or serpiginous distribution.

There are telangiectasias, which suggest a poikilodermatosis; In some cases, small, tan-red papillomas appear around the openings of the mouth, anus and vagina.

A sign that is almost pathognomonic, soft herniations of subcutaneous fat, yellow and saccular at the level mainly of the iliac crest and the inguinal and posterior region of the thigh. There is usually microcephaly and the hair is sparse and brittle. Nails absent or dystrophic, concave, grooved or hyperpigmented.

Ocular anomalies such as chorioretinal and iris colobomas, strabismus, nystagmus, tear duct obstruction, microphthalmia, and unilateral anophthalmia. Hypoplastic ears and deafness.

Congenital malformations such as bilateral syndactyly between the third and fourth fingers, clinodactyly, polydactyly, oligodactyly and adactyly, as well as brachydactyly.

There is mental and growth retardation, genital hypoplasia, dental defects, oligodontia, microdontia, enamel fragility.

Other reported malformations are cleft lip and palate, aortic stenosis, atrial septal defect, renal and ureteral anomalies.

Bone alterations are preferentially located in the limbs and are accompanied by syndactyly, oligodactyly and adactyly.
In some cases there is scoliosis, spina bifida and hypoplasia of the right clavicle, dystrophic nails, microdontia and coloboma of the iris.

Mental retardation in many of these patients.

Paraclinical examinations

Biopsy of fatty skin lesions. Normal adipose cells in the dermis. In scarred areas the changes are less specific.

Radiology. Striated osteopathy and the skeletal alterations already described.

Ultrasound. For diagnosis of malformations described.

Syndromogenesis and etiology

All the cases described are in females, which is why it is believed that focal dermal hypoplasia is incompatible with survival in male fetuses.

It is transmitted in a dominant way linked to the X chromosome, which is lethal in men. It is also possible that it is a dominant trait, but limited to sex. It is always accompanied by other ectodermal and mesodermal defects.

The etiology is genetic, probably monogenic.

Literature

Goltz, R.W. et al.: "Focal dermal hypoplasia syndrome. A review of the literature and report of two cases." Arch. Dermatol., 101:1-11, 1970.

GORLIN-GOLTZ SYNDROME

Synonymy

S. of cutaneousomandibular polyoncosis. Basal cell carcinoma.S. basal cell nevus.

Syndromography

Clinic

This syndrome is characterized by being hereditary and by presenting multiple defects in organs and systems of ectodermal and mesodermal origin that are described below:

1. Cutaneous. Basal cell epitheliomas; milia, epithelial and sebaceous cysts; lipomas; dyskeratosis of palms and soles.
2. Bones. Mandibular cysts; bifid (beveled) and synostotic rib; brachymetacarpalism; occult spina bifida; scoliosis; defective dentition.
3. White fabrics. Calcification in falx cerebri and elsewhere; ovarian fibromas with calcification; lymphatic cysts of the mesentery.
4. Nervous system. Mental and neuropsychic abnormalities; non-specific electroencephalographic alterations; agenesis of the corpus callosum.
5. Eyepieces. Hypertelorism; singing dystopia; cataracts; congenital amaurosis.

These patients are abnormally sensitive to radiotherapeutic doses of ionizing radiation, since patients treated in this way have developed a large number of basal cell tumors in the irradiated area shortly after exposure to the treatment.

Syndromogenesis and etiology

Of genetic etiology with a hypothesis similar to other neoplasms in which tumor suppressor genes have been found, it is transmitted as an autosomal dominant inheritance with complete penetrance and extreme variation in gene expressivity. 40% are due to new mutations in which advanced paternal age appears to be the fundamental cause.

Carcinomas fit Knudson's hit theory, the first hit being the germline mutation that is transmitted in an autosomal dominant pattern and the second hit being the action of mutagenic agents such as radiation.

Literature

Binkley, G.W. and H.H. Johnson, Jr.: "Adenoid Epithelioma Cysticum: Basal Cell Neci, Agenesis of the Corpus Callosm and Dental Cysts, A.M.A.". Arch. Dermat. Syph., 63:73, 1951.
Clendenning, M.E.; J.E. Block and L.C. Radde: "basal cell nevus syndrome." Arch. Dermat., 90:38-53, 1964.
Gorlin, R.J. and R.N. Goltz: "Multiple nevoid basal-cell epithelioma, jaw cysts and bifid ribs: Syndrome." New Eng. J. Med., 262:908, 1960.

Gorlin, R.J. and K.W. Goltz,: “Multiple nevoid basalcell carcinoma Syndrome.” Medicine, 66:98-113, 1987.
Katz, J.; R. Savin and H.M. Spiro: "The basal cell nevus syndrome and inflammatory disease of the bowel." Amer. J. Med., 44:483-488, 1968.

Neblett, CR. et al.: “Neurological involvement in the nevoid basal cell carcinoma syndrome.” J. Neurosurg., 35:577, 1971.
Nomland, R.: “Multiple basal cell epithelioma originating from congenital pigmented cell nerve.” Arch. Derm., 08, 1932.

GOUGEROT SYNDROME

Synonymy

Hailey-Hailey S. Benign chronic familial pemphigus S.
S. of hereditary bullous dyskeratosis.

Syndromography

Clinic

It is seen in both sexes and usually begins in adolescence or early adulthood, usually in warm, humid climates. It is characterized by the appearance of unilateral or bilateral lesions in friction areas (neck, armpit, groin) and sometimes other areas (skull, parts of a limb).

These lesions are in the form of clusters and are composed of small vesicles that initially contain a clear fluid that later becomes cloudy, accompanied by erythema of the surrounding skin. They later break and form scabs.
They can extend to the periphery.

Paraclinical examinations

Skin biopsy. Lesions similar to pemphigus vulgaris are observed, but it differs in the following: more extensive acantholysis, less damage to acantholic cells, presence and permanence of few intercellular bridges.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance and it has been suggested that the defect is due to an adhesion abnormality of keratinocytes that makes their union difficult.

Literature

Burge, S.M.; P.R. Millard and F. Wojnaroswka: “Hailey-Hailey disease: a widespread abnormality of cell adhesion.” Brit. J. Derm., 124:329-332, 1991.

Gougerot (1933) quoted by Gougerot, H.: "La priorité du pemphigus chronique familial héréditaire benin". Ann. Dermat. et Syph., 10:361-363. 1950.

Montgomery, H.: Dermatopathology. V. 1, New York, Harper Row, 1967.

GOUGEROT-CARTEAUD SYNDROME

Synonymy

S. of confluent reticular papillomatosis.

Syndromography

Clinic

It predominates in the female sex and begins at puberty. It is characterized by the appearance of flat papules no larger than 1.5 cm in diameter that are located between the breasts and in the midline of the back. The lesions spread, become confluent and form an irregular network that radiates in all directions from the neck to the pubis.

Paraclinical examinations

Skin biopsy. Hyperkeratosis and papillomatosis without tir acanthosis.

Syndromogenesis and etiology

These are generally isolated cases, but it is suspected that it is a genetic condition.

The etiology is unknown.

Literature

Gougerot, H. and A. Carteaud: "Papillomatose pigmentée innominée." Bull. French Soc. Dermat. Syph., 34:719-721, 1927.

Gougerot, H. and A. Carteaud: “Papillomatose pigmentée confluente et reticulée innominée”. Bull. French Soc. Dermat. Syph., 35:230, 1928.

Kesten, B.M. and H.D. James: "Pseudoatrophoderma colli, acanthosis nigricans, and confluent and reticular papillomatosis." Arch. Derm., 75:525-542, 1957.

GREITHER SYNDROME

Synonymy

S. de queeratosis palmaris et plantaris familiaris. S. of Greither's keratosis. Unna-Thost disease.

Syndromography

Clinic

This syndrome is observed in both sexes and begins in childhood. It is characterized by palmoplantar keratosis that progresses until the fourth decade of life; The lesions extend to the back, hands and feet, in the form of irregular patches on the arms and legs.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. Low serum vitamin A levels have been reported in some cases. He

Topical use of vitamin A obtains good results. It is considered that there are two clinical types: a common form and a severe one.

The latter of autosomal recessive inheritance.

The etiology is genetic and presents heterogeneity.

Literature

Gamborg Nielsen, P.: Two different clinical and genetic forms of hereditary palmoplantar keratoderma in the northernmost country of Sweeden". Clin. Genet., 28:361-366, 1985.

Greither, A.: "Keratosis extremitatum hereditaria progrediens mit dominam Erbagang." Hautarzt., 3:198-203, 1952.

GROENBLAD-STRANDBERG-TOURAINE SYNDROME

Synonymy

S. of Groenblad-Strandberg. S. of pseudoxanthoma elasticum.

S. of dystrophic elastosis. Systemic elastic disease.

S. of hereditary elastodystrophy. S. of generalized elastorhexis.

S. de Darier.

Syndromography

Clinic

This syndrome can manifest itself completely or partially. It can appear at any age. It is characterized by skin lesions in the form of bands or small yellowish papules, which are located in the stretch marks or grooves of the skin of the neck, armpits, flexion side of the elbows, and popliteal areas.

The skin is not elastic when pulled for a while, the opposite of Ehlers-Danlos syndrome. Eye injuries:

narrow reddish or gray lines around the papilla in both eyes.

Occasionally, degeneration of the retina and choroid.

Cardiovascular: alteration of peripheral pulses, angina pectoris, hypertension and sometimes visceral hemorrhages (hematemesis and melena), or symptoms of focal brain injury.

Paraclinical examinations

Skin biopsy. Degeneration and fragmentation of the elastic fibers of the skin. In addition, arteritis lesions in different organs.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance. Sometimes it is associated with the syndromes of: Paget, Marfan, Herrick, Albers Schönberg

The etiology is unknown.

Literature

Darier, J.: “Pseudoxanthoma elasticum.” Monatsh. Prakt. Dermat., 23:609-617, 1896.

Doyne (1889) by Geominne, L.; E. Van Ginneken and R. Bernard: "Groenblad-Strandberg-Touraine Syndrome." Press. Med., 71:2511-2514, 1963.

Groenblad, E.: “Angoid streaks: Pseudoxanthoma elasticum. Vorlaufige Mitteilung". Acta Ophthal., 7:329, 1929.

Strandberg, J.: "Pseudoxanthoma elasticum." Zentralbl. Haut. or. Gschl. Krth.. 31:689, 1929.

Touraine, A.: L'elastorrhexie systémataisée". Bul1. Soc. Franc. Dermat. et Syph., 47:255-273, 1940.

HABER SYNDROME

Synonymy

Familial rosacea-type rash with intraepidermal epithelioma.

Syndromography

Clinic

It is seen in both sexes and begins in childhood. It is characterized by a rash, resembling rosacea, on the nose, cheeks, forehead and beard. There is also erythema, telangiectasias, follicular papules with areas that resemble pitting. Later in life, non-symmetrical, scaly or keratotic verucous lesions 1 cm in diameter appear on the trunk and thighs.

Paraclinical examinations

Skin biopsy. Erythematous lesions show:

perivascular infiltration with fibrosis, acanthosis, parakeratosis and proliferation of sebaceous glands. Verrucous lesions show: papillomatosis, acanthosis, dyskeratosis and mitotic figures.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Sanderson, K.V. and H.T.H. Wilson: "Haber's syndrome-familial rosacea-like eruption with intraepithelial epithelioma." Brit. J. Dermat., 77:1-8, 1965.

Wilson, H.T.H.: Two cases of familial rosacea-like dermatosis with lanugo hair changes." Brit. J. Derm., 72:322, 1960.

HALLOPEAU II SYNDROME

Synonymy

S. of pyodermitis vegetans. S. of pemphigus vegetans (variety Hallopeau).

Syndromography

Clinic

It is observed in both sexes in middle age. It is characterized by the formation of pustules (non-flicthenous) followed by warty vegetations that open and erode. The most frequent location is in the flexor areas and mouth.

Paraclinical examinations

Skin biopsy. Non-specific granulomas with abscess formation, pseudoepitheliomatous hyperplasia, acanthosis and slight hyperkeratosis.

Syndromogenesis and etiology

It is considered a benign variant of pemphigus.

The etiology is unknown.

Literature

Hallopeau, H.: Nouvelle note sur la dermatose bulleuse hereditaire et traumatique". Arch. Derm. Syph., 45:323-328, 1898.

HELWEG-LARSEN SYNDROME

Synonymy

S. of anhidrosis-neurolabyrinthitis.

Syndromography

Clinic

It is seen in both sexes from birth. There are temperature disorders (fever) due to anhidrosis or marked hypohidrosis. In the fourth to fifth decades of life, vertigo due to neuro-labyrinthitis is associated with progressive perceptual hearing loss.

Physical examination. The skull, hair and teeth are of normal configuration. Dryness of the skin accompanied by follicular hyperkeratosis on the palms of the hands and feet.

Syndromogenesis and etiology

Three types of generalized anhidrosis are considered:

1. Ectodermal dysplasia with anomalies of teeth, hair and sweat glands.
2. Ectodermal dysplasia with no other abnormalities, and sweat glands with abnormal morphology and function.
3. Ectodermal dysplasia without other defects or sweat gland abnormalities.

In the syndrome described there is a congenital hypoplasia of the sweat and sebaceous glands, with hypohidrosis.

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Helweg-Larsen, H.F. and K. Ludvigsen: "Congenital familial hydrosis and neurolabyrinthitis." Acta Derm. Vener. Stockh., 26:489-505, 1946.

Lewkunia, R.M. and R.B. Lorry: "Progressive hemifacial atrophy (Parry-Rosemberg syndromes). Report with review of genetics and nosology." Am. J. Med. Genet., 14:385-390, 1983.

BULOUS ICHTHIOSIFORM HPERKERATOSIS SYNDROME

Syndromography

Clinic

It is observed in both sexes normally at birth or in the first 7 days. It is characterized by vesicular eruptions associated with erythema and peeling that appear quickly and recur. In the third month, lesions with a hyperkeratotic appearance appear that are more evident around the fourth month and more visible in the flexion areas. General health is good. Hair and nails are normal.

Paraclinical examinations

Skin biopsy. Hyperkeratosis and acanthosis. Perivascular inflammatory infiltrates.

Syndromogenesis and etiology

Abnormality of the stratum corneum due to reduced epidermal transit time increases moisture loss through the abnormal barrier.

It is transmitted by autosomal dominant inheritance. The eruptions decrease with age and are rare in adulthood. Only hyperkeratotic lesions persist.

Literature

Brocq, L.: “Erythrodermie congenitale ichthy osi form avec hyper epidermotrophy." Ann. Derm. Syph., 3:1, 1902.
Frost, P. et al.: "Icthyosiform dermatoses. III. Studies of transepidermal water loss." Arch. Dermatol., 98:230-233, 1968.

CONGENTTA VILLOUS HYPERTHRICOSIS SYNDROME

Synonymy

Hypertrichosis lanuginosa universalis. S. of trichostasis. S. of congenital hypertrichosis lanuginosa. S. of "boy with a dog face." S. of "werewolf. S. of Jansen-Cornelia de Lange.

Syndromography

Clinic

It is due to excessive and persistent development of fetal lanugo. Some children are born with hypertrichosis and in others it develops in the first years of life. It is characterized by the presence of silky hair that covers almost the entire body with the exception of the palms and soles. This hair can be 2-10 cm in length; The eyebrows are wide and bushy, they appear double. This hair is persistent.

It is associated with hypodontia or anodontia and gingival fibromatosis. Occasionally there are atrial malformations. Psychosomatic development is normal, with exceptions.

Syndromogenesis and etiology

The production mechanism is not well clarified, some think that it may be due to an arrest in skin development, since the lesions found are: lanugo follicles that remain long instead of atrophying as normal, but the hair bulbs are in the subcutis and not in the corium.

Etiology. The "benign" form is transmitted by autosomal dominant inheritance. There is another "malignant" form in which they are born with hypertrichosis, a simian appearance, thick lips and prognathism; its transmission is uncertain.

Literature

Felgenhauer, E.R.: Hypertrichosis lanuginosa universalis". J. Gen. Hum., 17:1, 1969.
Janssen, T.A.E. and C. de Lange: "Familial hypertrichosis totalis (Trichostasis)". Paediatr Act. (Uppsala), 33:69, 1945.

HYSTRIX ICHTHYOSIS SYNDROME

Syndromography

Clínica

It is observed in both sexes and generally begins in childhood. It is characterized by marked hyperkeratosis with hard, cone-shaped scales that rise in the shape of a hedgehog and envelop the entire body, with the exception of the face, palms of the hands, soles of the feet and genitals.

Paraclinical examinations

Skin biopsy. It shows extreme hyperkeratinization of the skin.

Syndromogenesis and etiology

It is considered an autosomal dominant hereditary transmission.

The basic defect is found at the level of the fibrillar structural protein of keratinocytes or tonofibrils that are formed in large quantities and are concentrated surrounding the nucleus in the form of shells; There is another type of ichthyosis hystrix gravior in which the tonofilaments are rudimentary.

Literature

Anton-Lamprecht, I.: “Electron microscopy in the early diagnosis of genetic disorders of the skin." Dermatologica, 157:65-85, 1978.
Curth, H.D. and M.T. Macklin: "The genetic basis of various types of ichthyosis in a family group". Am. J. Human. Genet., 6:371-381, 1954.
Penrose, L.S. and C. Stern: Reconsideration of the Lambert pedigree (icthyosis hystrix gravior)". Ann. Human Genet., 22:258-283, 1957.
Rook, W.; D.S. Wilkinson and F.J.G Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

LAMINAR ICHTHYOSIS SYNDROME

Synonymy

S. de Brocq. S. of congenital non-bullous ichthyosiform erythroderma.

Syndromography

Clinic

It occurs in both sexes and the lesions are present at birth. It is characterized by generalized erythema and large scales (5-15 mm) of brownish gray color, with a striking quadrilateral configuration, which are free at the edges and adhered in the center. In severe cases they are so thick that they are compared to a shell. These changes are most severe in the flexures and neck. There is a variety in which the lesions are more generalized, which is called collodion baby syndrome (see).

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

It should be clarified that the original Brocq syndrome included both the bullous and non-bullous forms. Today it is considered that they are different not only in their histological appearance, but also in their mode of transmission. For this reason, the majority of cases so far included in the non-bullous variety are those that belong to lamellar ichthyosis.

The etiology is genetic.

Literature

Brocq, L.: "Erythrodermis congenitale ichthyosiforme avec hyperepidermotrophie." Ann. Derm. Syph., 3:1-31, 1902.
Lenz, C.L. and J. Altman: “Lameliar ichthyosis". Arch. Derm., 97:3--13. 1968.

DOMINANT VULGAR ICHTHYOSIS SYNDROME

Synonymy

S. of xeroderma. S. of simple ichthyosis. S. of ichthyosis nitida.

Syndromography

Clinic

It is observed in both sexes between 1-4 years of age. It has no symptoms except for skin irritation with the cold.

Physical examination. Rough and dry skin in cold temperatures with small lesions and the stratum coneum cracks and produces rhomboid scales. These injuries progress over the years and become more severe. The affected areas are: the extension surfaces of the limbs and the trunk. The back has more severe injuries than the anterior part of the thorax and abdomen.
Armpits and other areas are spared. The face can be affected in children. Symptoms are exacerbated in winter and return in summer.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The basic defect has been linked to a biochemical abnormality of filaggrin, which may function with the matrix protein keratin of stratum coneum cells.

The etiology is genetic.

Literature

Felsher, Z. and S. Rothman: Insensible perspiration of skin in hyperkeratotic conditions." J. Invest. Dermat., 6:271-278, 1945.
Wells, R.S.: “Tchthyosis.” Brit. Med. J., 2:1504-1506, 1966.

SEX-LINKED VULGAR ICHTHYOSIS SYNDROME0

Synonymy

Ichthyosis nigricans S.

Syndromography

Clinic

It is only observed in men and begins in childhood. Injuries can cause disfigurement.

Physical examination. Disseminated lesions with brown scales that affect almost all areas of the skin, preferably the armpit, forearms, popliteal fossa, front part of the trunk more marked than the back. Tendency to disappear or decrease in spring and autumn. Alopecia may occur and may also be associated with mental retardation, skeletal abnormalities, and hypogonadism.

Paraclinical examinations

Skin biopsy. The granular layer thickened with acanthosis and lymphocytic infiltrates in the dermis.

Slit lamp examination. This eye exam shows clouding of the cornea.

Syndromogenesis and etiology

It is transmitted by sex-linked recessive inheritance and women do not suffer from the syndrome.

The basic defect consists of a deficiency of steroid sulfatase. The gene is located in Xp22-pter.

Literature

Kerr, C.B. and R.S. Wells: "Sex-linked ichthyosis." Ann. Hum. Genet.. 29:33-50, 1965.
Wells, R.S.: “Icthyosis.” Brit. Med. J., 2:1504-1506, 1966.
Badur, H.P. and J.C. Koedar: Disorders of keratinization in connective tissue and its Heritable disorders (Royce, P. and B. Eteinmann, eds.), New York, Wiley-Liss, 1993, pp 680-681.

JACQUET SYNDROME

Synonymy

S. of reflex alopecia. S. of congenital circumscribed alopecia.

Syndromography

Clinic

Circumscribed alopecia occurs in association with other ectodermal dysplasias that affect: nails and teeth (Hallerman Shreiff syndrome, pseudopelada syndrome and others), or associated with epidermal nevi (which is the most common form).
Alopecia can be present from birth or develop in the first month of life.

Syndromogenesis and etiology

It is considered to be of genetic origin, but its chthyology is unknown.

Literature

Jacquet, L.: "Des érythemes papuleux fessiers post-érosifs." Rev. de Mal. de Lénf., 4:208-218, 1886.
Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

JADASSOHN-PELLIZARI SYNDROME

Synonymy

S. of anetoderma. (See S. de Schweninger-Buzzi.)

Syndromography

Clínic

It predominates in women and generally begins in the second to fourth decades of life. It is usually located on the trunk, shoulders and face. It manifests itself with well-defined erythematous spots, the size of a lentil, which soon wither in the center and form a circular lesion, within which the epidermis is slightly wrinkled. Over time the lesion depresses. Later the surface becomes shiny, white and wrinkled. The examining finger clearly sees a hole, with defined edges as if it were an inguinal hernia. This is explained by the loss of elastic tissue at that level. There are no sensory disorders or alterations in other regions of the skin.

Paraclinical examinations

Skin biopsy. It shows a perivascular lymphocytic infiltrate in the dermis; But the fundamental alterations are based in the elastic tissue, whose fibers fragment and may even disappear completely.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Jadassohn, J.: "Ueber eine eigenartige Form von Atrophia maculosa cutis." Verhnadl. d. Dutch. Dermat. Gesellsch., 1891, pp. 342- -358.

Korting, G.W.; J. Cabré and H. Holzmann: Zur Kenntnis der Kollagenveranderungen bei der Anetodermie vom Typus Schweninger-Buzzi". Arch. Klin. Exp. Derm., 218:274-297, 1964.

Pellizari, C.: "Erythema orticato atrofizzante: atrophy parziale idiopatica della pelle." Gior. Italian. D. Mal. Ven., 19:230-243, 1884.

JESSNER-KANOF SYNDROME

Synonymy

S. of lymphocytic infiltration of the skin.

Syndromography

Clinic

Almost exclusively in men between the second and eighth decades of life. It is characterized by one or multiple flat discoid papules of pink or red-brown color, which subsequently grow to form raised plaques.

Sometimes these plates are circinate, but firm. Generally the molar region of the face is affected, as well as the eyelids and some other part of the body may show lesions. Sun exposure can irritate or aggravate the injury. No visceral or systemic signs of intake were found.

Paraclinical examinations

Skin biopsy. The epidermis is normal, but the dermis presents a diffuse lymphocytic infiltration that also covers the perivascular and adnexal regions of the skin.

Syndromogenesis and etiology

A specific etiology of this syndrome has not been demonstrated and in previous years it was considered a variety of chronic discoid lupus and by others a form of lymphocytoma of the cutis. (See S. de Spiegler-Fendt.)

Literature

Jessner, M. and N.B. Kanof: "Lymphocytic infiltration of the skin." Arch. Derm. Syph., 68:447-449, 1953.

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology, Philadelphia, Davis, 1968.

JOB SYNDROME

Synonymy

S. of Job's dermatitis. Variety of chronic granulomatous disease. Buckley's S.

Syndromography

Clinic

It occurs in women. It begins in the first year of life and is characterized by recurrent suppurative infections of the skin, which take on the appearance of cold, painless subcutaneous abscesses. Precisely for this reason it has been called Job syndrome, based on a biblical precept that says: “Satan He stigmatized Job with pimples, from the soles of his feet to the crown of his head." The cases studied are patients with white skin and red hair, with somewhat coarse facies and short stature. There is facial and flexure dermatitis, and onychodystrophy.

Cold lung abscesses, lymphadenitis and conjunctivitis may occur.

Paraclinical examinations

Blood. Type E hyperimmunoglobulinemia well above normal values (30-600 IU) and chemotaxic defect in neutrophils.

Syndromogenesis and etiology

This syndrome is transmitted by autosomal recessive inheritance. Some authors suggest three genetic hypotheses:
1, mosaicism for the trait; 2, homozygosity of the recessive gene; 3, dominance with variable expressiveness.

Abscesses are caused by staphylococcus aureus. This syndrome seems to be part of the spectrum of granulomatous diseases or syndromes.

The etiology is genetic and the basic defect lies in the chemotaxis of neutrophilic granulocytes and high levels of IgE in serum.

Literature

Bannatyne, R.M.; P.N. Skowron and J.L. Weber: "Job syndrome, a variant of chronic granulomatous disease". J. Pediatr., 75: 236, 1969.

Buckley, R.H.; BB Wary and E.Z. Belmaker: “Extreme hyperimmunglobulinemia E and undue susceptibility to infections." Pediatrics, 49:59, 1972.

Davis, S.D.; J. Schaller and R.J. Wedgewood: Job syndrome, recurrent 'cold' staphylococcal abscesses". Lancet, I:1073, 1966.

Quie, P.G.: "Chronic granulomatous disease of childhood." Advance Pediatr., 16:287, 1969.

KAPOSI SYNDROME

Synonymy

S. of idiopathic hemorrhagic sarcoma.

Syndromography

Clínic

Regarding women, it predominates in men (10:1).
It occurs most frequently in European Jews, Italians, and blacks. It begins in middle ages of life.

The first lesions appear on the arches of the forearms, hands, legs and feet in the form of blue-black, reddish or purple macules, which extend and converge to form large plaques or become nodules. These nodules have a hard or rubbery consistency and appear in the form of dark blue angiomas. At first there may be furfuraceous edema in the affected parts and later there is swelling with intense lymphedema during the disease. In more advanced periods the nodules appear on the face, ears, trunk and soft palate. These lesions are confused with granulomas, angiomas or neurofibromas.

Generally the lesions are progressive and when they subside they leave atrophic, depressed and hyperpigmented scars. There is an increase in lymph nodes and occasionally visceral lesions (liver and spleen). Sometimes it is associated with diabetes and lymphomas.

Paraclinical examinations

Radiology. Bone lesions are found such as: rarefaction, cysts and cortical erosion, which are characteristic, and demonstrate the dissemination of the tumor.

Biopsy of the lesion. Requires the diagnosis.

Syndromogenesis and etiology

In this syndrome, there is a proliferation of immature multipotent cells derived from the reticuloendothelial system, motile, which sometimes develop and transform into phagocytic cells and other times into giant cells and are related to lymphoblastomas; or they can be considered as an angiomatosis that arises from pluripotent vascular cells.

This syndrome is the most common neoplasm in patients with AIDS, which suggests that immunological deficiencies play an important role in its development.

The etiology is unknown.

Literature

Kaposi, M. and M. Kohn: “Idiopathisches, multiple Pigmentsar kom der Haut.” Arch. f. Dermat. or. Syph., 4:265-273, 1872.

KYRLE SYNDROME

Synonymy

S. of follicular and parafollicular hyperkeratosis penetrating the cutis.

Syndromography

Clinic

Age of onset between 20 and 63 years without difference in both sexes. This syndrome is a rare form of follicular disease in which the lesions are their own. There is a follicular hyperkeratosis that forms a horn that projects into the skin, so that when each one of them is extracted, a depression similar to a hole is left. Papules are usually small, but they may fuse to form circinate plaques. They are located, preferably, in the lower limbs, but sometimes they occur in other parts of the body, excluding the mucous membranes and the palms of the hands and soles of the feet;

When the lesions evolve, atrophic scars remain.

Paraclinical examinations

Skin biopsy. Large keratotic spikes are observed in the follicles and sweat ducts and also in other areas. These plugs penetrate through the dermis and cause an inflammatory and foreign body giant cell reaction around the lower end of the plug in the dermis.

Syndromogenesis and etiology

Kyrle syndrome is especially associated with diabetes, but also with liver or kidney conditions. It has been suggested that it is a genodermatosis with abnormal carbohydrate metabolism that is inherited in an autosomal recessive manner. An abnormal metabolism of vitamin Ay is also thought to occur in an infectious process.

The etiology is unknown.

Literature

Carter, V.H. and V.S. Costantine: "Kyrle's disease I'. Arch. Derm., 97:624-632. 1968.

Kyrle, J.: "Hyperkeratosis follicularis et parafollicularis in cutem penetrans." Arch. Derm. Syph., 123:466-493, 1916.

Costantine, V.S. and V.H. Carter: "Kyrle's disease II." Arch. Derm., 97:633-639, 1968.

LASSUEUR-GRAHAM-ITTLE SYNDROME

Synonymy

Graham-Little S. S. of Brocq's pseudopelada. S. of lichen spinulose.

Syndromography

Clínic

It is observed in both sexes and at all ages. It is characterized by scarring alopecia of the skull, trunk and limbs with follicular plugging and keratosis pilaris.
Occasionally, alopecia of the axilla, pubis and trunk is observed.

Syndromogenesis and etiology

Possible development variant. It tends to be considered a variety of follicular lichen planus.

The etiology is unknown.

Literature

Brocq, L.: *Alopecia". J. Cut. Ven. Dis., 3:49-51, 1885.

Graham-Little, E.G.: “Folliculitis decalvans et Atrophicans”. Brit. J. Derm., 27:183-185, 1915.

Waldorf, D.S.: “Lichen planapilaris.” Arch. Derm., 93:684-691, 1966.

LEINER SYNDROME

Synonymy

S. of desquamative erythroderma.

Syndromography

Clinic

It predominates in the female sex. It occurs in children between 2-4 months old. The onset is rapid and is characterized by severe seborrheic dermatitis on the skull, and flexures, with macules and/or plaques or erythema with peeling on the trunk and limbs. At first there is a diffuse inflammatory redness that covers the entire body, which later becomes covered with grayish-white scales, which can be fine and furfuraceous or very large, and exfoliate into true sheets. As the process develops, the exfoliation can be general, with cracking and thickening of the skin, the nails are destroyed; the scalp has a thick crust. Digestive disorders, fever and mild lymphadenopathy may appear.

Paraclinical examinations

Skin biopsy. It shows seborrheic dermatitis with an added infection caused by Candida species (66%) and staphylococci.

Syndromogenesis and etiology

Infectious factors, already mentioned, and a decrease in complement C5 have been considered; metabolic alteration due to a biotin deficiency.

The etiology is unknown.

Literature

Leiner, C.: "Erytrodermia Desquamativa (universal dermatitis of children at the breast)." Brit. J. Dis. Child., 5:244-251, 1908.

PROFUSE LENTIGINOSIS SYNDROME

Synonymy

Multiple lentiginosis S. S. of lentiginosis. S. of the leopard. S. cardiocutaneous. S. of cardiomyopathic lentiginosis. S. of lentigo-electrocardiographic changes.

Syndromography

Clínic

This syndrome is observed in both sexes. Clinical findings may be partial or total.

The appearance resembles leopard skin. In this syndrome, lentigo is formed by dark brown macules 1-5 mm in diameter, which are preferably located on the trunk, although other areas may be affected, such as the palms of the hands and soles of the feet, the oral mucosa, the genitals and the scalp. The lesions appear shortly after birth and have a characteristic mottled appearance that has given rise to the name leopard syndrome. Other anomalies may coexist: cardiac (electrocardiographic conduction defects, pulmonary valve stenosis with systolic murmur at the base), genital (gonadal hypoplasia, hypospadias, abdominal testes, delayed puberty), growth retardation, sensorineural deafness, pterygium colli. Familial profound blindness, ocular hypertelorism. There may be skeletal alterations such as pectus carinatus and/or pectus excavatum, kyphosis, winged scapula and mandibular prognathism.

Psychological development is normal.

Paraclinical examinations

Radiology. Hypertrophic cardiomyopathy, discrete or atypical pulmonary valve stenosis, skeletal alterations indicated in the symptoms are also demonstrated, as well as hyperflexibility of the metacarpophalangeal joints and zygodactyly.

Syndromogenesis and etiology

This syndrome is transmitted by autosomal dominant inheritance with high penetrance and variable expression.

The etiology is monogenic genetics.

Literature

Gorlin, R.J. et al.: “Multiple lentigines syndrome.” Am. J. Dis. Child., Matha: OD2, 1969, N.L.: "Lentigo and electrocardiographic changes." New Eng. J. Med., 278:780-781, 1968.
Pickering, D. et al. : Little Leopard Syndrome": Description of 3 cases and review of 24". Arch. Dis. Chil., 46:85, 1971.
Sommer, A. et al.: “A family study of the leopard syndrome". Am. J. Dis., Child., 121:520, 1971.
Walther, R.J.; B.J. Polansky and I.A. Grotis: “Electrocardiographic abnormalities in family with generalized lentigo." New Eng. J. Med., 275: 1220-1225, 1966.

LINDBERG SYNDROME

Synonymy

S. of cutaneous polyarteritis nodosa with livedo and nodules.

Syndromography

Clinic

It is observed in both sexes and at all ages. It is characterized by a rash in the form of nodules on the skin of the feet, legs and thighs; less frequently on the trunk, face and back. The lesions are generally distributed along the vessels and are small, reddish and sometimes painful. Nocturnal myalgias are common. Livedo may precede or follow the appearance of nodules.

Paraclinical examinations

Skin biopsy. Necrotizing arteritis of the subcutaneous vessels is observed. Fibrinoid degeneration, neutrophil infiltration.

Syndromogenesis and etiology

The etiology is unknown. (See S. de Kussmaul Maier:)

Literature

Lindberg, K.: *Ein Beitrag zur Kenntnis der Periarteritis nodosa". Acta Med. Scand., 76:183-225, 1931.

LOFGREN SYNDROME

Synonymy

S. de Besnier-Boeck-Schaumann.

Syndromography

Clinic

It is common in Irish, Scandinavian and Puerto Rican women and occurs during pregnancy or the postpartum period. It is characterized by fever and signs of bilateral enlargement of the mediastinal lymph nodes.

Paraclinical examinations

Radiology. Bilateral mediastinal lymph nodes are confirmed.

Kweim reaction. It is generally positive. Tuberculin test: negative.

Node and liver biopsies. They are essential to establish the diagnosis.

Syndromogenesis and etiology

Many cases are due to sarcoidosis. It has also been observed in Hodgkin's disease and viral infections. It has been considered a variety of sarcoidosis.

Literature

Lofgren, S.: "Erythema nodosum; studies on etiology and pathogenesis in 185 adult cases." Acta Med. Scand. (suppl.), 174:1-197, 1946.

LOHUIZEN SYNDROME

Synonymy

S. de cutis marmorata telangiectatica. S. of generalized congenital phlebectasia.

Syndromography

Clinic

It begins at birth and is characterized by a network of dilated veins in the skin, which can be localized in certain regions or generalized, and which are associated with spider veins and small ulcers.

Syndromogenesis and etiology

The etiology appears to be genetic. Several evidences of autosomal recessive inheritance have been reported.

Literature

Kurczynski, T.W.: "Hereditary cutis marmorata telangiectasia congenita." Pediatrics, 70:52-53, 1982.

Mizrahi, A.M. and P.M. Sachs: "Generalized congenital phlebectasia, report of a case." Amer. J. Dis. Child., 112:72-75, 1966.

Van Lohuizen, C.H.J.: "Uber eine seltene angeborene Hautanomalie (Cutis Marmorata Telangiectatica Congenita)". Acta Dermatovener, 3:202-211, 1922.

LORTAT-JACOB-DEGOS SYNDROME

Synonymy

S. of benign mucosal pemphigoid. S. of mucosynechial ocular pemphigus. Variety of Duhring-Brock, with conjunctival adhesions.

Syndromography

Clinic

It predominates in women 2:1 in relation to men and usually begins around 65 years of age.

It is characterized by recurrent blisters on the mucosa and skin adjacent to body orifices. They are more common in the conjunctiva and oral mucosa. The conjunctivae are affected in 75% of patients. These vesicles eventually rupture and leave erosions that take time to heal. At the conjunctiva they can leave fibrous adhesions (symblepharon). Also present are: xerosis, entropion, trichiasis and corneal opacities. Finally, the adhesions join both eyelids to the ocular globe and narrow the palpebral opening. The associated scarring can develop successive attacks of inflammation, vesicles and denudation of the mouth, pharynx, esophagus, nose, glans and vagina. Stricture of the esophagus may occur.

In one third of patients, tight skin blisters similar to bullous pemphigoid occur. Generally, the general condition is not affected much, but it tends to create blindness due to conjunctival lesions.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Lortat-Jacob, E.: "Benign mucosal pemphigoid; dermatite bulleuse muco-synéchante et atrophiante." Brit. J. Dermat., 70:361-367, 1958.

LUPUS ERYTHEMATOSUS SYNDROME

Syndromography

Clínic

It predominates in women of reproductive age. It presents with general malaise, asthenia, anorexia, migratory joint pain, abdominal pain and/or intermittent pleuritic pain.

Anaphylactoid pneumonitis" characterized by tachypnea and cyanosis. Mental retardation, anxiety, hallucinations, convulsions and recurrent febrile symptoms. Exposure to sunlight can induce the manifestations.

Physical examination. Butterfly wing rash in the nasal and malar region in 50% of cases. Erythematous maculopapular rash on the neck and limbs. Telangiectasias, chronic ulcers of the limbs, scars on the nail beds and ends of the fingers. Retinal exudates in cotton flakes.

Adenomegaly, splenomegaly and hepatomegaly. Signs of cardiovascular disorders (pericarditis, endomyocarditis in 50% of cases) and kidney disorders in 75% of patients. As seen, it is a systemic condition that can affect any "white" tissue, although one of its main characteristics is the lesion on butterfly wings already mentioned.

Paraclinical examinations

Blood. Leukopenia, eosinopenia, anemia, thrombocytopenia, hypergammaglobulinemia, presence of " lupus anticoagulant." LE positive cells.

Accelerated erythrocyte sedimentation. Positive serology for syphilis for a long time before the appearance of symptoms should raise suspicion of the condition.

Urine. Albuminuria.

Syndromogenesis and etiology

It is considered produced by immunological mechanisms. Congenital susceptibility is suspected. The origin seems to be due to the rupture of cells that release antigens. These antigens bind with anti-DNA and are phagocytosed resulting in the release of lysosomal products. They in turn cause the destruction of new cells and the cycle reproduces.

In this way, autoantibodies and pathogenic immune complexes are produced that are associated with an inability of the humoral and cellular immune system to quickly get rid of these products and also to suppress their production. Vasculitis and lupus anticoagulant" are the cause of visceral lesions, which explains the symptoms of this syndrome.

In lymphocytes from patients with lupus, stimulated by phytohemoagglutinin, the synthesis of two types of DNA occurs: a high molecular weight fraction like that of the controls and an excess of low molecular weight DNA not found in the controls. The excess of low molecular weight DNA has been found to be independent of drugs such as procainamide (drug induction was suggested in lupus, as an expression of pharmacogenetic polymorphism). But the expression of excess low-weight DNA appears to have an autosomal dominant inheritance. On the other hand, excess low-weight DNA precedes the appearance of antinuclear antibodies, which may represent a predisposing factor.

Other research suggests that the increase in oxidative metabolism in lymphocytes from patients stimulated with phytohemoagglutinin can generate chemical changes in the DNA.

endogenous 'in vivo' and this, in turn, may be a primary event in the pathogenesis of autoimmunity observed in patients with systemic lupus erythematosus.

Familial aggregation and the above studies carried out in patients and relatives suggest an autosomal dominant inheritance.

Literature

Leucovitz, H. et al.: "Segregation of lymphocyte low-molecular weight DNA and antinuclear antibodies in a family with systemic lupus erythematosus in first cousins." Huma. Genet., 80:253-258, 1988.

LYELL SYNDROME

Synonymy

S. of burned skin. S. of erythroderma bullosa with epidermolysis. S. of acute febrile toxic epidermal necrolysis pemphigoid. S. of drug dermatitis.

Syndromography

Clinic

It is observed in all ages. It is characterized by skin irritability, pain and fever. Some cases present systemic manifestations that can be of small or great intensity with signs of toxicity and shock. It can cause lesions in different organs, mainly liver, pancreas and intestine (colon). The first case in the small intestine was reported in 1994 by P. Michel and collaborators in France.

Physical examination. Erythema that begins around the mouth and in a few hours invades the entire body followed by large blisters. Moderate pressure on the skin produces peeling of the epidermis and leaves reddish areas of variable size.

Paraclinical examinations

Skin biopsy. Epidermal necrosis is observed without signs of vascular damage.

Syndromogenesis and etiology

They may be reactions to different drugs, especially in adults, or be in association with staphylococcal infections in children. (See Ritter syndrome:)

The etiology is unknown.

Literature

Debre, R.; M. Lamy and M. Lamotte: "Un cas dérythrodermie avec epidermolyse chez un enfant de 12 ans." Bull. Soc. Pediat. 37:231--238, 1939.

Delchier, J.C.; 0. Chosidow: *Atteinte intestinale au cours du syndrome de Lyell, à propos de 4 cas". Gastroenterol. Clin. Biol., 15, A25, 1991.

Lyell, A.: "Toxic epidermal necrolysis (the scalded skin syndrome)." Br. J. Dermatol., 100:69-86, 1979.

MAJOCCHI SYNDROME

Synonymy

S. of telangiectoid annular purpura.

Syndromography

Clinic

It is observed in both sexes and generally begins in adolescence or young adulthood. Anywhere on the body the skin

It may be affected by small annular plaques (1-3 cm in diameter) of purple or yellow-tan color. They extend eccentrically and may present an area of atrophy in the center of the lesion.

Paraclinical examinations

Skin biopsy. Demonstrates telangiectasias and hemosiderin deposition.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Majocchi, D.; A. Rook; D.S. Wilkinson and FJ.G Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

MANTOUX SYNDROME

Synonymy

S. of ulcer-hypertension

Syndromography

Clínica

It affects both sexes and generally begins in youth. It is characterized by a rash on the palms of the hands, soles of the feet and fingers of translucent miliary papules that grow slowly and form a dark central area. They last for a few weeks and leave small spots that may disappear later. They tend to be recurring for years and sometimes for short periods.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Mantoux, C.: “Porokératose pokillomateuse palmaire et plantaire", Ann. by Derm. et Syph., 4:15-31, 1903.

MARTORELL SYNDROME

Synonymy

S. of ulcer-hypertension

Syndromography

Clinic

It predominates in middle-aged women. It is characterized by very painful ulcerations of the limbs, frequently bilateral, located above the knees. In addition, the characteristic symptoms of high blood pressure.

Syndromogenesis and etiology

It is due to high blood pressure. In these cases, histology has shown accumulation of a hyaline material between the elastic layer and the endothelium.

Literature

Martorell, E.: Hypertensive ulcer. Barcelona Ed., B.T.P., 1953.

MELEDA SYNDROME

Synonymy

Meleda's disease. S. of Mljet. S. of palmoplantar hyperkeratosis. S. of palmoplantar tylosis. S. of mutilating plamoplantar keratosis.

Syndromography

Clinic

It is seen in both sexes and begins in the first months of life. It is characterized by redness of the palms of the hands and soles of the feet followed by localized or diffuse fine peeling that progressively extends across the dorsal surface. The erythema remains. It is accompanied by hyperhidrosis and eczematization, often in the form of patches and new lesions on the limbs.

Diffuse, symmetrical keratinization of the skin that begins in the palmoplantar regions and sometimes extends to the dorsum of those regions. Produces keratomas. There is hyperhidrosis and maceration. In severe forms, self-amputation may occur.

The term keratoderma is frequently used synonymously with keratoma, hyperkeratosis, keratosis and tylosis.

Mental development is poor and delayed.

Paraclinical examinations

Skin biopsy. Marked acanthosis is found;
hyperkeratosis and parakeratosis of irregular distribution and lymphohistiocytic infiltrates in the perivascular regions.

Syndromogenesis and etiology

Changes in the cornification of the palms of the hands and soles of the feet, with ichthyosiform changes in different regions, characterize this disease, which receives this name due to its relatively high frequency per inhabitant on the island of Meleda in the Mediterranean Sea.

It is due to excessive keratin formation. There are two varieties:

Acquired. Climacteric keratoderma and grooved plantar keratoma. It is also observed in other diseases (chronic eczema, psoriasis, porokeratosis, etc.).
congenital It is what concerns us. Keratoderma can be seen in other congenital conditions:
ichthyosis, Papillon-Lefèvre syndrome, Touraine polykeratosis and Meleda syndrome.

In general, the direct cause of keratoderma is the excessive formation of keratin in the palms of the hands and soles of the feet.

It should be noted that Franceschetti and Schnyder have carried out a clinicogenetic classification of hereditary keratoderma.

It is transmitted by autosomal dominant inheritance.

The etiology is genetic.

Literature

Franceschetti, A. and U.V. Schnyder: “Attempt at clinical genetic classification of hereditary palmo plantar keratoses whit consideration of associated symptoms". Dermatologica, 120:154-178. 1960.
Islamon, T. and 0. Lazovic: "Contribution á l'étude de la maladie de Mljet." J. Genet. Hum., 10:172-201, 1961.
Kogoj, F.: "Die Krankheit von Mljet ("Mal de Meleda"). Acta Dermatovener. Stockh., 15:264-299, 1934.
Touraine, A.: "Essai de Classification des Keratoses Congenitales". Ann. Derm. Syph., 85:257, 1958,

MENDES DA COSTA II SYNDROME

Synonymy

S. of variable erythrokeratoderma. S. of progressive and symmetrical congenital erythrokeratoderma. S. of progressive erythrokeratoderma.

Syndromography

Clinic

It occurs in both sexes and begins in the early years, before the age of 3. Rarely later. It is characterized by the appearance of circumscribed erythematous and hyperkeratotic plaques anywhere on the body. The plates can take on a variegated geographical-looking configuration and are clearly delimited. In a short time, hours or days, they change configuration or disappear completely. The areas most frequently affected are the extension surfaces of the limbs, buttocks and face.

Paraclinical examinations

Skin biopsy. Edema, cellular infiltration, hyperkeratosis and acanthosis are seen to a variable degree.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. Two clinical forms have been reported: one called erythroderma variabilis and another progressive asymmetric. Both appear to be due to dominant mutations, although it has been suggested that they are different manifestations of a single entity.

The etiology is genetic.

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MacFarlane, A.W.: S.J. Chapman and J.Z. Verbod: "Is erythrokeratoderma one disorder? A clinical ultrastructural study of two siblings." Br. J. Derm. , 124:487-491, 1991.
Mendes da Costa, S.: "Ery throdermia and keratodermia variabilis in mother and daughter." Acta Dermatovener.. 6:255-261, 1925.

MIESCHER SYNDROME

Synonymy

S. of disciform granuloma. Macular necrobiosis S.
S. de Miescher-Leder.

Syndromography

Clinic

It predominates in women between 50-75 years of age, although it has been reported in adolescents. It is characterized by marginalized reddish-yellowish plaques, generally polycyclic in shape with

an atrophic center. These formations are indurated and are located on the dorsal surface of the hands, forearms and anterior part of the legs. The abdominal and thoracic wall may also be taken.

Paraclinical examinations

Kweim test.

Skin biopsy. Granulomatous infiltration with giant cells; These lesions begin around the vessels and end up obliterating them.

Syndromogenesis and etiology

It is a very controversial syndrome, in some cases sarcoidosis has been confirmed, hence the value of performing the Kweim test to eliminate this condition.

Literature

Miescher, G.: “Spotted nekrobiosis.” Dermatologica, 98:199-204, 1949.

MORPHEA SYNDROME

Synonymy

S. of localized morphea S. of generalized scleroderma.

Syndromography

Clinic

More common in women than in men in a 3:1 ratio and generally begins in the second to fourth decades. It is characterized by migraines, arthralgias, abdominal pain in 15% of patients. Joint pain in 40% of cases.

Physical examination. 5 varieties have been considered:

1. Plates, usually multiple, asymmetrical (2-15 cm in diameter); areas of induration of the skin that in a few months become smooth, shiny, without hair or sweaty secretion. They are ivory in color. Vesicles, hemorrhages and telangiectasias may develop. There is hypoesthesia in these areas. It can affect the trunk, face and genital areas.

2 Larger and less numerous lesions, similar to white spot syndrome (see) .

3. Linear lesion generally simple, sometimes bilateral. The injury, although linear, is the same as the injuries described previously.

4. Frontoparietal scleroderma, same as coupe de sabre syndrome. The linear scleroderma lesion affects the frontal and parietal regions of the face and skull and is associated to a lesser or greater extent with subcutaneous atrophic changes.

5. Large widespread plaques that appear on the trunk and sometimes all over the body. The face loses its expression and there are contractures.

Paraclinical examinations

Skin biopsy. The epidermis is normal or atrophic. The dermis was edematous with collagen degeneration and moderate perivascular lymphocytic infiltration. There is a reduction in elastic fibers, atrophy of hair follicles and sweat glands.

Radiology. Spinal abnormalities in 47% of cases.

Syndromogenesis and etiology

Sometimes, at the beginning it is associated with trauma, pregnancy or menopause, but no obvious relationship has been established. It has been shown that there is no increase in collagen fibers, but rather an increase in hexosamines linked to collagen.

The etiology is unknown.

Literature

Dilley, J.J. and H.O. Perry: Bilateral linear scleroderma in coupe de sabre." Arch. Derm., 97:688-689, 1968.

Fagge, C.H.: “On keloid scleriasis, morphoca and some allied affections.” Guy's Hosp. Rep., 13:255-328, 1868.

MANY-HABERMAN SYNDROME

Synonymy

S. of pityriasis lichenoides. Glutatic parapsoriasis S. S. of varioliform parapsoriasis. S. de Wise.

Syndromography

Clinic

It predominates in men and generally begins in adolescence and young adulthood, rarely in children. In adults it takes the chronic form and in children the acute form.

Acute form. It is generally preceded by mild systemic manifestations for 2-3 days: fever, general malaise, headache, inflammation of the joints with arthralgia, rash. Later, papular, cdematous, reddish or reddish-brownish lesions with vesicles in their center and hemorrhagic necrosis around them begin suddenly, leaving a crusty scar ulceration. Vesicular lesions are not common, but they are of great diagnostic value and are located deeply. The papulonecrotic lesions with brown-blackish scabs and hemorrhagic excoriations are very striking. Then Cura's exanthema appears, leaving uniform, pigmented, depressed and varioliform scars.

The usual location is in the anterior part of the trunk, the flexion surfaces of the upper limbs and the armpits.

The palms of the hands or soles of the feet or the mucous membranes are not affected.

Chronic form. Small brown lichenoid papules are present. Scabs are not frequent nor is peeling.

The condition lasts from 4 weeks to 6 months and in addition to the scars, temporary leukodermic plaques can sometimes be seen in the two forms described. Some cases have recurrent seizures for years.

Paraclinical examinations

Skin biopsy. It varies with the stage of the injuries. At the beginning, lymphocytic infiltration, capillary dilation and endothelial proliferation, as well as necrotic changes, can be seen.
In the chronic stages they are very similar to the lesions produced by eczema or psoriasis, which is why some have considered it a form of parapsoriasis.

Syndromogenesis and etiology

Essentially it is a vasculitis whose etiology can be viral or allergic.

Literature

Habermann, R.: "Ueber die akut velaufende, nekrotisier ende unterart der Pityriasis lichenoides lichenoides (Pityriasis lichenoides et variolformes acuta)." Derm. Zsch., 45:42-48, 1925.
Mucha, V.: "Uber einen der Parakeratosis variegata (Unna) bzw. Pityriasis lichenoides chronica (Neisser-Juliusberg) nahestehen den eigentumlichen Fall". Arch. F. Dermat. u Syph., 123:586- -592, 1916.

NAEGELI SYNDROME

Synonymy

S. of melanophoric nevus. S. de Franceschetti-Jodassohn.

S. of skin pigmentation-hyperhidrosis-keratosis pilaris-dysphasia.

Syndromography

Clinic

It is observed in both sexXOS in the second to third years of life. It is characterized by the development of reticular pigmentation in the form of a fine network that extends over several areas without previous inflammatory changes. Keratoderma of the palms of the hands and soles generally exists.

Body temperature regulation may be altered due to a reduction in the number of sweat glands. Hair and nails are normal. The teeth are normal or with some defects consisting of yellow spots.

Paraclinical examinations

Dermatoglyphs. Fingerprint hypoplasia.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. It is very rare, it has been described only in Swiss families.

The etiology is monogenic genetics.

Literature

Franceschetti, A. and W. Jodassohn: «"A propos de l'incontinentia pigmenti' délimitation deux syndromes différents figurants sous le meme terme". Dermatologica, 108:1-28, 1954.
Naegeli, O.: Familiarer Chromatophorenaevus". Schweiz. Med. Wschr., 57:48, 1927.

SUBCUTANEOUS NODULAR FATTY NECROSIS SYNDROME

Synonymy

S. of liquefactive nodular panniculitis.

Syndromography

Clínica

The lesions are characteristic and differ from those of classic erythema nodosum; they vary in size from a few millimeters to centimeters, and in contrast to erythema nodosum, they are mobile. They are red and painful, evolve in 2-3 weeks and can leave a hyperpigmented, slightly depressed scar. The nodules frequently coincide with abdominal pain and may be accompanied by arthralgia and fever.

The lesions are usually located on the legs and rarely involve other regions. The largest nodules are They become fluctuating and become abscess-like, rupture and exude a whitish, creamy or oily viscous material. This is exceptional in erythema nodosum.

Syndromogenesis and etiology

This syndrome can be associated with acute pancreatitis and also with neoplasms of the pancreas, especially adenocarcinoma.

NETHERTON SYNDROME

Syndromography

Clinic

It occurs only in girls and is characterized by congenital ichthyosiform erythroderma or linear ichthyosis with associated bamboo hairs. Banmbo hairs are observed not only on the scalp, but also on the eyebrows, eyelashes and, more rarely, in other areas. The lightening of the hair can be seen throughout the body. Hairs on bamboo can disappear in a few years.

There are important atopic manifestations.

Paraclinical examinations

Blood. Hypergammaglobulinemia has been reported.

Syndromogenesis and etiology

It has been suggested that it is transmitted by autosomal recessive, or dominant, inheritance with variable penetrance, although this has not been definitively confirmed.

The reported consanguinity shows more evidence of autosomal recessive inheritance; the possibility of an inborn error of metabolism has even been raised.

The etiology is monogenic genetics.

Literature

Netherton, E.W.: “A unique case of trichorrhexis nodosa: bamboo hairs.” Arch. Derm., 78:483-487, 1958.
Wilkinson, R.D.; G.H. Curtis and A. Hawk: Netherton's disease: trichorrhexis invaginata (bamboo hair) congenital icthyosiformi erytroderma and the atopic diathesis. "A his pathology study." Arch. Derm., 89:46-54, 1964.

NEUMANN SYNDROME

Synonymy

Pemphigus vegetans S., Neumann variety.

Syndromography

Clinic

It occurs in both sexes, in young people and in middle age. It is characterized by the appearance of flaccid blisters that rupture and form fungoid vegetations or papillomatous proliferations, especially in the intertriginous areas. Although the beginning of the lesions is sometimes manifested only by the bursting of the blisters in the mouth, genitals, perineum, armpits, groin, limbs, scalp and navel, the points of imperfect rupture of the same and their moist bases become exuberant, with warty vegetations that later constitute the main feature of the eruption; They are covered with scabs and surrounded by an inflammatory area.

The subjective symptoms are mild but sometimes a high fever and other symptoms due to sepsis develop, and the end is similar to pemphigus vulgaris with some intercurrent illness, for which there is a greater predisposition due to exhaustion and steroid therapy.

Paraclinical examinations

Skin biopsy. The characteristic findings are:

acantholysis, formation of intraepidermal cracks and blisters and the presence of acantholytic cells (Tzanck cells covering the blister and loose within the cavity). There is also papillary proliferation and marked epidermal hyperplasia, which differentiates this syndrome from pemphigus vulgaris, which presents lesions similar to those described above.

Syndromogenesis and etiology

Although the etiology is unknown, it has begun to be considered a phenomenon of autoimmunity, since intercellular autoantibodies have been found that are located in the areas of acantholysis in the intraepidermal region and their titer increases with the severity of the condition.

Literature

Neumann, I.: "Ueber Pemphigus vegetans (Frambosioides)". Vrtljschr. F. Dermat., 13:157-178, 1886.

ACHROMATIC NEVUS 0 ANEMCOUS SYNDROME

Syndromography

Clínic

It is observed in both sexXOs. It begins after birth or in the first years of life. Unilateral or bilateral lesion, characterized by the lack of pigment that takes the form of rounded, well-defined spots with irregular edges, sometimes as a linear lesion. They can appear in any location. Its distribution, texture and appearance do not change during life, nor are there any alterations in sensitivity in these areas. The border is not pigmented nor are there other cutaneous or systemic lesions.

Paraclinical examinations

Skin biopsy. Absence of melanophores.

Syndromogenesis and etiology

It has been described in four generations of a family, following an autosomal dominant inheritance pattern with reduced penetrance.

It is considered a congenital alteration of the mesenchyme, which inhibits the migration of melanoblast precursor cells.

The etiology is genetic.

Literature

Cardoso, H.; R. Vignale and H. Abreu de Sastre: “Familial naevus anemicus:. (Abstract)y". Am. J. Hum. Genet., 27:24A, 1975.
Coupe, R.L.: Unilateral systematized achromic naevus". Dermatologica (Basel)., 134:19-35, 1967.
Lesser, E.: In Ziemssen, H.V.: Handbook der Hautkrankeiten Bd 2, Leizig, Vogol., 1884, p. 183.

OCULOORGENITAL SYNDROME

Synonymy

S. orogenital. S. Jacobs.

Syndromography

Clínica

It is characterized by a burning sensation and intense itching in the scrotum, conjunctivitis, stomatitis and fissure of the wing of the nose.

Physical examination. There is scrotal dermatitis, pigmentation with scaling and superficial ulcerations.

Paraclinical examinations

Blood. Anemia and signs of malnutrition (hypoalbuminemia)

Syndromogenesis and etiology

It is due to a deficiency of riboflavin and other B complex vitamins. Among the different causes that can cause this deficiency is prolonged treatment with antibiotics.
Local inflammation added to lesions plays an important role in dermatopathy.

Literature

Gilbert, C.E: "Giant condyloma acuminatum (Buschke-Loewenstein tumor)." Arch. Derm. (Chicago), 93:714-717, 1966.
Jacobs, E.C.: "Oculo-oro-genital syndrome: A deficiency disease." Ann. Int. Med., 35:1049-1054, 1951.

PAPILLON-LEFÈVRE SYNDROME

Synonymy

S. of palmoplantar hyperkeratosis-premature periodontal destruction of the teeth. S. of familial palmar and plantar keratosis (tylosis). S. de Brünauer. S. of familial palmoplantar keratoderma:.

Syndromography

Clinic

It begins in the first 2-4 months of life, with skin and dental manifestations. It is characterized by palmoplantar hyperkeratosis that most frequently adopts the diffuse type, or sometimes the punctate type; generally this lesion is not severe. The plantar injury can go beyond the calcaneus and reach the Achilles tendon and the external malleolar and tibial tuberosities ("sandal"). However, in some cases it can be severe, especially in winter, and decrease or disappear in summer. There is foul hyperhidrosis especially in the feet. Simultaneously, hyperkeratosis appears, the gums become red, edematous and bleeding, and there is halitosis.

There is also destruction of the periodontal ligaments with pus formation, which causes the teeth to become loose, move easily and become detached. Then it attacks the permanent teeth and the process is repeated with the exception of the third molars.

In severe cases, total tooth loss occurs at 6-10 years of age. Treatment with acitretin, the free acid form of etretinate, applied early, has such good effects that patients develop normal teeth.

There may be systolic murmurs, adenoinfarctions and delay mental.

Paraclinical examinations

Radiology. It demonstrates ectopic calcifications in the tentorium and choroid layer of the eye.

Blood. Occasionally hyperglycemia and high hemoglobin levels.

Syndromogenesis and etiology

The lesions are very similar to those of Meleda disease.

It is transmitted by autosomal recessive inheritance and sometimes there is consanguinity of the parents.

The etiology is monogenic genetics.

Literature

Brünauer, St. R.: Zur Vererbung des Keratoma hereditarium palmare et plantare". Acta dermat.-venerol., 4:389, 1923.

Brünauer, St. R.: "Zur Symptomatologie und Histologie der Kongenitalen Dyskeratosen." Dermat. Zschr., 42:6-26, 1925.

Gorlin, R.J.; H. Sedano and V.E. Anderson: The syndrome of palmar-plantar hyperkeratosis and premature periodontal destruction of the teeth. A clinical and genetic analysis of Papillon-Lefèvre syndrome", J. Pediatr., 65:895, 1964.

Naik, D.N., A. Velau: G. Alavandar et al.: Papillon-Lefèvre syndrome". Oral. Surg., 25:19-23, 1968.

Papillon, M.M. and P. Lefèvre: "Deux cas de keratodernmia palmaire symetrique familiale (maladie de Meleda) chez le frere et la soeur: coexistence dans le deux cas d'alterations dentaires grave. Bull. Soc. Fran. Dermat. Syph., 31:82, 1924.

CONGENITA PACHYONOCULA SYNDROME

Synonymy

S. de Jadassohn-Lewandowsky.

Syndromography

Clínica

This syndrome is characterized by:

1. Marked thickening of the nails, which are very thick and increase towards the edges. The nail plates are very hard and firmly attached to the bed; This is occupied by a yellow, cone-shaped, keratotic mass that causes the nail to rise.
2. Blisters and palmoplantar keratosis.
3. Follicular papules located on the buttocks and limbs, grayish-black in color, with crater-shaped depressions that are filled by cone cones.
4. Leukoplakia of the oral mucosa.
5. Dyskeratosis of the conea and cataracts.

Syndromogenesis and etiology

It is transmitted in an autosomal dominant hereditary manner.

The etiology is unknown.

Literature

Merklen, EP. and A. Guillard: "Cas isolé de pachyonychie congenitale avec hyperkératoses cutanée et leucoplasiforme (syndrome du Jadassohn-Lewandowsky type Richl), varieté de lapolykératose congenitale de A. Touraine". Presse Med., 62:410, 1954.

PASINI SYNDROME

Synonymy

Albopapuloid epidermolysis bullosa.

Syndromography

Clinic

It occurs in both sexes and begins in young people or adults. It is characterized by small, white, firm papules, located perifollicularly and growing slowly up to 15 mm, which appear on the trunk, especially in the lumbosacral region. It is a variety of hyperplastic epidermolysis bullosa.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. It has been suggested that the COL 7Al gene may be a candidate for this mutation.

Literature

Pasini, A.: "Dystrophy cutaneobolloso-atrofizzante ed albo-papuloid". Gior. Ital. de Dermat. e Sifil., 69:558-564, 1928.

Ryynanen, M. et al.: "Human type VII collagen genetic linkage of the gene (COLT 7A1) on chromosome 3 to dominant dystrophic epidermolysis bullosa." Am. J. Human Genetic, 49:797-803, 1971.

PAUTRIER-WORINGER SYNDROME

Synonymy

S. of lipomelanotic reticulosis. S. of dermopathic lymphadenitis.

Syndromnography

Clinic

It is characterized by generalized erythroderma, diffuse blackish pigmentation of the skin (melanoderma) and lymphadenopathy.
This syndrome occurs associated with several diseases in which generalized erythroderma has become chronic. It can therefore accompany malignant lymphoma, generalized neurodermatitis, prurigo and other erythrodermas of unknown etiology. It frequently presents intense itching.

Paraclinical examinations

Skin and lymph node biopsies. On the skin, signs common to other dermatoses with which it is associated are observed.
The ganglia show: at the level of the pulp, proliferation of reticular cells that contain melanin and lipids. The lymphoid follicles are infiltrated by eosinophils, neutrophils and plasma cells and exhibit a simple granulomatous hyperplasia. If the erythroderma is of lymphomatous origin, the lymph nodes show the changes specific to this condition.

Syndromogenesis and etiology

It is the one that corresponds to the dermatosis with which it is associated;

It must be differentiated from malignant processes, Hodgkin's disease, Brill-Simmerx disease, etc. The stimulation of melanocytes by inflammation and itching produces the deposition of melanin in the skin and lymph nodes.

The etiology is unknown.

Literature

Pautrier, L.M. and F. Woringer: Note preliminaire sur un tableau histologique particulier de lésions ganglionnaires accompagnant des eruptions dermatologiques généralisées prurigineuses des types cliniques différents". Bull. Soc. Fr. Derm. Syph., 39:947-955, 1932.

Pautrier, L.M. and E Woringer: La Reticculose Lipomélanique". Ann. Derm. Syph., 8:257, 1937.

Pautrier, L.M. and F. Woringer: "Contribution a l' etude de l'histo- physiologie cutanée; á propos d'un aspect histopathologique nouveau du ganglion lymphaatique; la réticulouse lipo-mélanique accompagnant certaines dermatoses généralisées; les échanges entre la peau et le ganglion." Ann. Dermat. et Syph., 8:257-273, 1937.

Schnyder, U.W. and C.G Schirrer: "So-called, lipomelanotic reticulosis' of Pautrier-Woringer". Arch. Dermat. Syph., 70: 155-165, 1954.

JUVENILE PEMPHIGODE SYNDROME

Syndromography

Clinic

More common in males before 5 years of age. The onset is acute with discrete systemic symptoms. It is characterized by tense, clear-looking blisters, measuring 1-2 cm, on healthy skin on the face, lower abdomen, buttocks, and less frequently on the limbs. Pruritus is rare. The blisters persist for a long time.

After dissolution they heal quickly. There may be hyperpigmentation but it does not leave a scar. It is rarely located in the mucous

Paraclinical examinations

Blood. Eosinophilia 5-10%.

Skin biopsy. It demonstrates that the bullae or vesicles are located at the dermoepithelial junction, but the epidermis is intact.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Rook, A.; D.S. Wilkinson and F.J.B. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

PENFHIGUS VULGARIS SYNDROME

Syndromography

Clinic

It occurs in both sexes in middle age (40-60 years).

At first the lesions are located in the mouth in the form of blisters that break and form ulcers, as well as on other mucous membranes (vagina); Other times it starts in the English, face, neck, scalp and armpits. The fluid inside the blisters is clear at first, but later becomes hemorrhagic or even seropurulent. These blisters are tight but soon They become flaccid and rupture, giving rise to ulcers and bloody, exudative surfaces that bleed easily. The bloody surfaces soon become covered, in part, by scabs that have little or no tendency to heal and that extend over large surfaces by confluence with each other.

Healed lesions leave hyperpigmented plaques with little or no scarring. Nikolsky's sign is positive (exaggerated separability of the outer epidermal layer due to slight friction, as in pemphigus).

Paraclinical examinations

Skin biopsy. Suprabasal acantholysis is observed, with intraepidermal blisters that unfold the layers of the skin. There are acantholytic cells.

Syndromogenesis and etiology

It is attributed to a virus or an autoimmune type lesion, since autoantibodies have been detected in the cells of the lesions.

The etiology is unknown.

Literature

Anderson, H. J. et al.: "Pemphigus and other diseases." Arch. Derm., 101:538-546, 1970.

PERNIO SYNDROME

Syndromography

Clinic

This syndrome includes the syndromes of: acute chilblains, chronic chilblains, erythrocyanosis, lupus pernio, winter dermatitis, cold immersion feet, names by which it is also known.

Acute chilblains. They are more frequent in children and women; Generally these lesions are bilateral and symmetrical. There is a blue-reddish dermatitis with slight edema. Painful and burning sensations in parts exposed to very cold temperatures: fingers, toes, and legs. The acute stage lasts about a week and a brownish color appears that persists for months. Occasionally, there may be hemorrhagic reaction and ulcerations with or without infection, as complications.

Chronic chilblains. If the previous episodes are repeated, chronic lesions occur with residual fibrosis and atrophy of the skin and subcutaneous cellular tissue. During the hot season the lesions can disappear almost completely.

Paraclinical examinations

Biopsy of the lesions. Angiphitis with necrosis of the adipose tissue and chronic inflammatory reaction of the subcutaneous tissues is observed. If the lesions are chronic, there may be iron deposits that give hyperpigmentation to those areas.

Syndromogenesis and etiology

It is due to a reaction of the peripheral vessels to the cold. It is a slow freezing variety.

Literature

Miller, W.: De Pernionibus. Jence, Paris, 1680.

Allen, E.V.; N.W. Barker and E.A. Hines: Peripheral Vascular Diseases. ed. 3. Saunders. Philadelphia, 1962.

PETGES-CLÉJAT SYNDROME

Synonymy

S. of poikilodermatomyositis. S. de Petges-Jakovi. S. of atrophic vascular poikiloderma.

Syndromography

Clinic

Generally, it occurs in young adults. At the beginning, there is muscle weakness and mottled skin with hyperpigmentation and hypopigmentation, telangiectasias and atrophy. The skin changes resemble radiodermatitis:
later there may be ulcerations. The skin in these places is dry, furfuraceous and wrinkles like a cigarette paper. There may be intense pruritus and calcinosis. It is considered that this syndrome may be a manifestation of other skin diseases, such as mycosis fungoides, plaque parapsoriasis, lupus erythematosus, and chronic atrophying acrodermatitis.
There is muscle wasting.

Paraclinical examinations

Skin biopsy. In the beginning lesions there is slight infiltration of lymphocytes in the epidermis, dilation of capillaries and increase or loss of melanin pigment. Later, lesions similar to those of lupus occur (except for a minor degree of keratotic plugs) and atrophy of the epidermis with alterations of the elastic tissue that finally disappear.
Urine. High creatinuria.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Guy, W.: R.C. Grauer and FM. Jacob: Poikilodermatomyosiotis". Arch. Derm. Syph., 40:876-878, 1939.
Petges, G. and C. Cléjat: "Sclérose atrophique de la peau et myosite généralisée." Arch. Dermat. Syph., 7:550-568, 1906.

STAINED LEGS SYNDROME

Syndromography

Clínica

It is observed in diabetic patients between 50-60 years old, many of whom show evidence of neuropathies and/or retinopathies. On the skin, lesions can be seen in greater or lesser numbers, round or oval, circumscribed, located bilaterally or asymmetrically and superficially covered by pigmented atrophic skin. The lesions are not painful nor do they ulcerate. They are distributed over the anterior, lateral and middle regions of the legs below the knees. Patients do not report traumas or ignore them and do not draw the doctor's attention to this information. There may be hypoesthesia and decreased or absent reflexes.

Paraclinical examinations

Skin biopsy. Atrophy of the epidermis, discrete fibrosis;
separation of collagen fibers from the skin; pigmentation due to hemosiderin and increase in melanin due to atrophy.

Syndromogenesis and etiology

It is thought that this syndrome is due to microvascular manifestations of diabetes and that there may be trauma as a triggering element.

The etiology is unknown.

Literature

Melin, H.: *An atrophic circumscribed skin lesion in the lower extremities of diabetes". Acta Med. Scand., 176(Suppl. 423):1-75, 1964.
Murphy, R.A.: "Skin lesions in diabetic patients: the 'spottedleg' syndrome." Lahey Clin. Found. Bull., 14:10-14, 1965.

BESNIER PRURIGUS SYNDROME

Syndromography

Clinic

It is characterized by localized itching over the lesions, often associated with asthma. Both are exacerbated alternatively. The lesions are patches located on the knees, face, elbows and wrists.

Syndromogenesis and etiology

It is part of the atopic erythroderma syndrome. Generally this type of lesion follows childhood dermatitis.
The etiology is unknown.

Literature

Besnier, E.: “Premiere note et preliminaire observation pour servir D'Introduction a L'Etude des Prurigos diasthesiques". Ann. Derm. Syph., 3:634-648, 1892.
French, H.: French's Index of Differential Diagnosis. ed. 9, Williams & Wilkins, Baltimore, 1967.

PSEUDOPELADA SYNDROME

Syndromography

Clinic

The presence of this syndrome in sex and age varies with the etiology. In idiopathic cases it occurs in women in the third to fourth decades of life. The onset is insidious and is characterized by the formation of small rounded, smooth, soft areas with inflammatory signs (only moderate erythema) that extend and converge. The most affected region is the vertex of the head. In the areas of the lesion there are no hairs or they are isolated. These bald areas can be small or large and distributed or grouped. They have a white or pinkish color, with a smooth, shiny, atrophic surface (onion skin), similar to marble or ivory.

Paraclinical examinations

Skin biopsy. Lymphocytic infiltrates are observed around the vessels and hair follicles, followed by atrophy of the follicles and sebaceous glands. Sweat glands are isolated.

Literature

Degos, R.; R. Rabut, B. Duperrat et al.: "L'état pseudopeladique; réflexions á propos de cent cas d'alopéciescica-tricielles en aires, dápparence primitive du type pseudo-pelade". Ann. Dermat. et Syph., 81:5-26, 1954.

PRURIGINOUS PURPURA SYNDROME

Synonymy

Disseminated pruritic angiodermatitis.

Syndromography

Clinic

This syndrome predominates in males, begins in adulthood, occurs very frequently in spring and summer, and is characterized by itching around the ankles that then spreads to the rest of the legs and moves up the body, with the exception of the palms of the hands and soles of the feet.

The lesions that cause this pruritus consist of punctate red-orange purpuric macules that develop completely and can converge within 2 weeks. Malleolar edema is also observed. Chronic pruritus produces lichenification.

Paraclinical examinations

Skin biopsy. A perivascular inflammatory reaction is seen in the upper corium with edema of the vascular endothelium and infiltration of lymphocyte cells, histiocytes and red blood cells.

Syndromogenesis and etiology

The etiology is unknown.

A microbial cause is suspected.

Literature

Davis, E.: "Schoenlein-Henoch syndrome of vascular purpura." Blood, 3:129-136, 1948.

CLIMATERIC KERATODERMA SYNDROME Synonymy

S. of Haxthausen.

Syndromography

Clinic

It is seen in climacteric women and is characterized by palmar and plantar hyperkeratosis, especially on the edge of the heels where discrete, well-defined, regular, round or oval lesions appear, varying in size between a lentil and a chickpea. Eczematous complications are common. Generally there is obesity and high blood pressure and chronic arthritis.

Paraclinical examinations

Skin biopsy. Histologically, there is extensive parakeratosis but without microabscesses that are characteristic of psoriasis, a state which it frequently resembles clinically.

Syndromogenesis and etiology

Its etiology is unknown.

Literature

Haxthausen, H.: 'Keratoderma climacterium'. Brit. J. Derm. Syph., 46:161-167, 1934.

RENDU-OSLER-WEBER SYNDROME

Synonymy

S. of hereditary hemorrhagic telangiectasia. Osler's disease. S. of hemorrhagic angiomatosis. Familial recurrent epistaxis with telangiectasia. S. de Rendu-Osler. S. of Babington.

S. de Goldstein.

Syndromography

Clínic

It is a familial vascular anomaly that occurs in both sexes and is characterized by small bunches of dilated capillaries, distributed superficially over the mucous membranes and skin. These slightly raised lesions are located mainly on the lips, tongue, palate, nasal mucosa, ears, palms and soles, fingertips and nail beds. It is also located in different mucous membranes of digestive, renal, neurological organs, etc.

Generally, they cause bleeding of greater or lesser intensity that depends on the site of location: epistaxis, melena, hematemesis, hematuria, etc.

It affects both sexes equally. There has been epistaxis since childhood. Generally, it begins in adolescence. As the patient develops, the tendency to hemorrhage may decrease or worsen and be fatal. In middle age there is a development of telangiectasias and angiomas whose number increases with age. At puberty they first appear below the surface of the tongue and on the floor of the mouth.

Physical examination. During the second or third decade of life, circumscribed punctate lesions, bluish-red capillary and venous telangicectasias, and spider veins may appear on the skin and mucous membranes. A pulmonary aneurysm syndrome may occur with clubbing, cyanosis, polyglobulia, and other signs.

Paraclinical examinations

Study of coagulation. Normal except if associated with VonWillebrand syndrome. Hypochromic anemia if there has been bleeding.

Biopsy of a lesion.

Syndromogenesis and etiology

It is transmitted by simple autosomal dominant inheritance. It is considered to be caused by a physiological weakness in the contractility of the small terminal arterioles; as well as the insufficiency of the muscular and elastic tissues of the vessels that prevent a normal hemostatic process.

Hemorrhages are caused by erosions or ruptures of the dilated capillaries typical of the syndrome.

It is transmitted by autosomal dominant inheritance.

The etiology is genetic.

Literature

Garland, H.G and S.T. Anning: Hereditary hemorrhagic telangiectasia: a genetic and bibliographic study". Br. J. Derm., 62:289, 1931.

Osler, W.: "On a family form of recurring epistaxis associated with multiple telangiectases of the skin and mucous membranes." Bull. John Hopkins Hosp., 12:333, 1901

Rendú, M.: "Epistaxis repetées chez un subject porteur de petits angiomes cutanés et muqueux." Bull. Soc. Med., 13:731, 1896.

Weber, F.P.: “Multiple hereditary developmental angiomata (telangiectases) of the skin and mucous membranes associated with recurring hemorrhages.” Lancet, 2: 160, 1907.

Weber, FP.: "Hemorrhagic telangiectasia of the Oslertype 'telangiectatic dysplasia' and isolated case, with discussion on multiple pulsating stellate telangiectases and other stricking stricking telangiectases and other stricking haemangiectases conditions." Br. J. Dermat., 48:182-193, 1936.

SWEAT RETENTION SYNDROME

Synonymy

S. of anhydrotic tropical asthenia.

Syndromography

Clinic

It is characterized by: anhidrosis, miliaria, pruritic crisis, severe systemic symptoms, collapse.

Syndromogenesis and etiology

Fischer and Maidbach have described some cases related to neurological problems and think that it is due to lesions that affect the spinal decussation of the efferent vegetative fibers and also those that affect the preganglionic sympathetic nerves.

The etiology is unknown.

MULTICENTRIC RETICULOHISTIOCYTOSIS SYNDROME

Synonymy

S. of generalized giant cell histiocytoma. S. of reticulohistiocytic granuloma. S. of pseudosarcomatous reticulohistiocytoma. S. of lipoid rheumatism. S. de Nicolau--Balus.

Syndromography

Clinic

Predominant in the female sex (3:1), it occurs in all races and generally begins between the second and sixth decades.

The development is gradual and is characterized by nodules of the skin, mucosa, subcutaneous tissue, synovium, periosteum and bones that cause deforming polyarthritis. In 25%, nodules and arthritis appear simultaneously; In 60%, arthritis precedes the nodules by months or years, and more rarely it is the other way around. Other symptoms: weight loss, pruritus, asthenia, fever and paresthesias; 40% have xanthelasma; 27% hypertension and 16% lymphadenopathy.

Paraclinical examinations

Blood. Hyperlipidemia that reaches all fractions. Increased betalipoprotein.

Radiology. Shows bone lesions.

Skin biopsy. The nodules show vacuolated histiocytes that obstruct small blood vessels, with associated lymphocytic infiltration. There are multinucleated giant cells with lipid vacuoles inside that move the nucleus towards the periphery of the cell.

Syndromogenesis and etiology

Multicentric reticulohistiocytosis can be classified among the granulomatous syndromes with lipid deposition (see histiocytosis X syndrome, and Farber syndrome.)

There is another entity called Crosti syndrome and which is characterized by tumor infiltrations located in the posterior part of the body. The lesions contain histiocytes and large xanthomatous cells.

It can be classified among granulomatous syndromes.

This entity of unknown etiology must be differentiated from multicentric histiocytoma.

Literature

Cainelli, T.: “Reticulo-istiocytoma of the dorsum of the Di Crosti adult.” G.Ital. Derm., 150:505-5 12, 1964.

Davies, N.E.J.; H.H. Roenigk; W.A. Hawk, et al.: "Multicentric reticulohistiocytosis, report of a case with histochemical studies." Arch. Derm., 97:543-547, 1968.

Parkes-Weber, F. and W. Freudenthal: “Nodular nondiabetic cutaneous xanthomatosis with hyper-cholesterolemia and typical histological features". Proc. Roy. Soc. Med., 30:522-526, 1937.

Targett, J.H.: “Giant cell tumors of the integuments.” Tr. Path. Soc. (London), 48:230-255, 1897.

RIEHL SYNDROME

Syndromography

Clinic

More common in females and begins at any age.

It is characterized by gray-brownish pigmentation of the face, more intense on the forehead and temples. Occasionally it extends to the neck and skull, rarely to other parts. There is often itching and redness, especially at the beginning. Once spread, the spots remain stationary and melanosis develops slowly over several months. When there is pigmentation in the covered parts, it is located in areas exposed to friction such as the anterior axillary folds and the navel. There is diffuse hyperkeratosis that gives the skin the appearance of being "dusted with flour."

Syndromogenesis and etiology

The etiology is unknown, but there is evidence that it may be due to photosensitivity to sunlight in the presence or absence of perfumes, creams or some other substance, in predisposed subjects.

Literature

Riehl, G.: "Ueber ine eigenartige Melanose Vorlaufige Mitterung." Vienna. Klin. Wschr., 30:780-781, 1917.

ROTHMANN-MAKAL SYNDROME

Synonymy

S. of Makal.

Syndromography

Clinic

It predominates in children and affects both sexes equally. It is characterized by the appearance of nodules or plaques in the subcutaneous tissue, which are located symmetrically; They are firm, elastic in consistency and measure 10-15 cm in diameter. Its preferred site is the trunk and occasionally the lips and face and anterior parts of the thigh. It is not accompanied by systemic symptoms.

Paraclinical examinations

Skin biopsy. At the beginning, vasodilation, necrosis of fat cells and neutrophilic infiltration are seen.

In the intermediate period: histiocytes with foam cells. In terminal phase: fibrosis with lobar atrophy.

Syndromogenesis and etiology

It is considered that it may be due to trauma or vascular injuries.

The etiology is unknown.

Literature

Laymon, C.W. and W.C. Parson, Jr.: Subcutaneous lipogranulomatosis (Rothmann-Makal), an appraisal." Arch. Derm., 90:288-292, 1964.

Makal, F: Uber Subcutaneous Lipogranulomatosis." Klin, Wschr., 7:2343-2346, 1928.

Rothmann, M.: "Ueber Entzundung und Atrophie des subcutanen Fettgebes." Virchow Arch. Path. Anat. Phys., 136:159-169, 1894.

ROTHMUND-THOMSON SYNDROME

Synonymy

S. de Rothmund-Werner (association of S. de Rothmund with S. de Werner). S. of congenital-atrophic-vascular poikiloderma. S. of mesoectodermal dysplasia with bone disorders.

S. de Bloch-Stauffer.

Syndromography

Clinic

It generally occurs in females and begins with tense, pink plaques on the cheeks, hands, feet and buttocks, which are followed by fine or punctate reticulated atrophy, associated with telangiectasias and brown pigmentation. These signs appear between 3-6 months of life. There is frequently short stature, small hands, absence or lightening of eyebrows and eyelashes, alopecia of the scalp and congenital bone defects. There may be hypogonadism, dystrophic nails and defects in teeth. Juvenile cataracts appear in more than 50% of patients between 3 and 6 years of age.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance. Abnormalities in the karyotypes of fibroblasts from areas of poikiloderma have been reported, suggesting instability.

chromosomal in vitro.

The etiology is genetic.

Literature

Der Kaloustian, V.M. et al.: "Clonal lines of aneuploid cells in Rothmund-Thompson syndrome." Am. J. Med. Genet., 37:336- -339, 1990.

Rothmund, A.: "Uber Cataracten in Verbindung mit einer eigennthumlichen Hautdegeneration." Graefe. Arch. Ophthal., 14:159-182, 1968.

Silver, H.K.: *Rothmund-Thomson syndrome: an oculocutaneous disorder:". Amer. J. Dis. Child., 111:182-190, 1966.

RUD SYNDROME

Synonymy

S. of male hypogonadism and ichthyosis.

Syndromography

Clinic

It occurs in both sexes in childhood. It is characterized by ichthyosiform erythroderma, acanthosis nigricans, dwarfism, mental deficiency, epilepsy, hypogonadism and chronic anemia and retinitis pigmentosa.

Paraclinical examinations

Hemogram. Megaloblastic type anemia.

Syndromogenesis and etiology

The etiology is monogenic genetic, it is transmitted as a recessive inheritance linked to the X chromosome with a male-female ratio of affected individuals of 2:1 due to manifestations in heterozygotes. The link between Kallermann syndrome and X-linked ichthyosis has suggested the possibility of reported families suffering from a continuous gene syndrome. The affected man does not reproduce.

Literature

Rud, E.: "Et Tilfaelde af Infantilisms med Tetani, Epilepsy, Polyneuritis, Ichthiosis og Anaemi of Pernicios type." Hospitalstidene, 70:525-538, 1927.

Wisniewski, K.; A.R. Levis and A.L. Shamke: “X-linked inheritance of the Rud syndrome". (Abstract) Am. J. Human Genet., 37:A83, 1985.

York-More, M.E. and A.T. Rundle: “Rud's syndrome.” J. Ment. Def. Res., 6:108-118. 1962.

DE SANCTIS-CACCHIONE SYNDROME

Synonymy

S. pigmentary xeroderma with neurological component and malignant tumor. S. of xerodermiaidiocia.

Syndromography

Clinic

It is observed in both sexes and begins after birth or in the first years of life. It is characterized by lentiginous lesions and areas of pigmentation that are related to sun exposure and increase with age.

Squamous cell neoplasms or carcinomas are common and more rarely melanomas.

It may have a clinical picture with manifestations in different organs or systems.

Neurological disorders: language disorders, mental deficit, spastic paralysis and seizures. These disorders are associated with microcephaly. Eye disorders: photophobia, keratitis, ectropion. Endocrine disorders: growth retardation, gonadal hypoplasia.

Paraclinical examinations

Skin biopsy. It may show alterations in pigmentation and basal cells, with a tendency to degeneration.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

The basic defect is found at the level of DNA repair enzymes similar to XP type A (xeroderma pigmentosa). In this syndrome the repair rate of DNA exposed to ultraviolet light is less than 10%. The neurological manifestations are related to the fact that neuronal death can occur more quickly in people with defects in DNA repair enzymes.

Literature

De Sanctis, C. and A. Cacchione: L'idiozia xerodermica". Riv. Spe. Freniat. 54:269-292, 1932.

Lynch, H.T.; OF. Anderson; J.L. Smith, Jr, et al.: "Xeroderma pigmentosum, malignant melanoma, and congenital ichthyosis." Arch. Derm., 96:625-635, 1967.

Kanda, T. et al.: "Peripheral neuropathy in xeroderma pigmentosum." Brain, 113:1025-1044, 1990.

Pick, FJ.: "Ueber Melanosis lenticularis progressiva." Vschr. Derm. Syph., 11:3-32, 1884.

SAVILL SYNDROME

Synonymy

S. of epidemic exfoliative dermatitis.

Syndromography

Clínic

This is an epidemic generally limited to closed communities, for example: institutions. Exfoliative dermatitis affects the face, skull, arms, rarely trunk and legs. Eccematous lesions may be added.

Syndromogenesis and etiology

It is very similar to infectious eczematoid dermatitis.

(See Sulzberger-Garbe syndrome.)

The etiology is unknown.

Literature

Savill, T.: "On an epidemic skin disease." Br. Med. J., 2:1197- -1202, 1891.

SCHAFER SYNDROME

Syndromography

Clinic

It is characterized by a disseminated follicular keratosis of the skin, with leukokeratosis of the oral mucosa; small foci of alopecia, dysonychia, congenital cataract, microcephaly, mental retardation, hypogonadism and dwarfism.

Literature

Silver, H.K.: "Rothmund-Thompson-syndrome: An oculocutaneous disorder", Amer. J. Dis. Child., 111:182-190, 1966.

SCHAMBERG SYNDROME

Synonymy

S. of progressive pigmented purpuric dermatosis.

Syndromography

Clinic

It is more common in males and occurs at any age. The initial lesions are like reddish dots the size of the head of a pin, they appear in groups that combine and constitute irregularly formed plaques, which progress slowly outward due to the development of fresh lesions in the periphery. After a few months the color of the spots fades, and they become brownish or yellowish; They simulate peppercorns on the skin. There are no subjective symptoms and it is rare that the affected area can recover its normal appearance over time. It is located with a predilection on the tibial crests and ankles.

Paraclinical examinations

Skin biopsy. A diffuse infiltrate of the subpapillary layer is seen. Dilated vessels with swollen endothelia and hyaline degeneration of the wall are also observed, as well as hemosiderin deposits, which provide color to the pigmented areas.

Syndromogenesis and etiology

Stasis can play an important role. Family cases have been reported.

The etiology is unknown.

Literature

Schamberg, J.F: “A peculiar progressive pigmentary disease of the skin.” Br. J. Dermat., 13:1-5, 1901.

SCHWENINGER-BUZZI SYNDROME

Synonymy

S. of anetoderma. (See S. de Jadassohn-Pellizzary.)

Syndromography

Clinic

It predominates almost exclusively in the female sex and appears between the third and fifth decades of life.

It begins with lesions under the skin, in the form of nummular bladders, bluish-white, with a slate tinge, 1-2 cm in diameter, some of which have fine telangiectasias passing over them. Clinically there are no alterations in the skin that covers them. By applying pressure with the tip of a finger, almost all injuries can be reversed and the finger then penetrates a hole that offers little resistance; almost the same as what occurs in Jadassohn anetoderma. These lesions also mostly occur on the trunk. Progress is slow and is maintained by the appearance of new lesions as the old ones spontaneously regress, leaving soft, depressed scars.

Paraclinical examinations

Skin biopsy. No inflammatory changes are found.

There is fragmentation and disappearance of elastic fibers with thickening of collagen fibers.

Syndromogenesis and etiology

It is worth clarifying that anetoderma means atrophy and laxity of the skin and is due to the loss of elastic fibers, without any other important skin alteration being observed. Muscle atrophy has given rise to much confusion, since its manifestations are not very well defined. Two syndromes that have similar lesions have been described: Jadassohn-Pellizzary and the one we describe.

The etiology is unknown.

Literature

Korting, G.W.; J. Cabré and H. Holzmann: "Zur Anetodermie von Typus Schweninger-Buzzi". Arch. Klin. Exp. Derm., 218:274- -297, 1964.

Schweninger, E. and s. Buzzi: Multiple benign geschwulstartige Bildungen der Haut Internat. Atlas seltener Hautkrankht. Heft.
V. Hamburg u. Leipzig, Voss, 1891.

SENEAR-USHER SYNDROME

Synonymy

S. of pemphigus erythematosus.

Syndromography

Clínica

It is characterized by the appearance of blisters on normal skin that then rupture and leave crusty lesions similar to seborrheic dermatitis. They are usually located on the nose, cheeks (on butterfly wings). Also in the ears, that is, in places similar to those found in lupus erythematosus. The lesions are erythematous, hence their name, and sometimes hyperkeratotic. These lesions can be seen on the trunk, chest and limbs. It is a comparatively benign type of chronic pemphigus in which health is maintained. Sometimes it becomes pemphigus vulgaris or foliaceous.

Paraclinical examinations

Skin biopsy. Similar to pemphigus vulgaris, but the acantholytic alterations affect the most superficial strata of the epidermis (stratum granulosum). Antibodies have been found in the intercellular spaces and in the basement membrane, adding another link between lupus erythematosus and this condition.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Montgomery, H.: Dermatopathology. New York, Hoeber, 1966, p. 148.
Senear, F. and B. Usher: “An unusual type of pemphigus; combining features of lupus erythematosus." Arch. Derm. & Syph., 13:761- -781, 1926.

SÉZARY SYNDROME

Synonymy

S. of Sézary's reticulosis. Malignant reticulemic S.

Syndromography

Clinic

It is characterized by generalized erythroderma, splenomegaly, and superficial lymphadenopathy; Skin changes are accompanied by intense itching.

The skin shows generalized erythroderma of a typical bright red color, with leonine facies, eyelid edema, ectropion, and diffuse alopecia. There is also palmoplantar hyperkeratosis and dystrophic nails. Intense itching with burning, fever, chills and profuse sweating is very common. In addition, lymphadenopathy is usually found in the neck, armpits and groin.

Paraclinical examinations

Blood. Leukocytosis between 6,000 and 30,000. Atypical cells are observed in the circulating blood, lymph nodes, and skin infiltrate, which contain a large, convoluted nucleus, with a thin ribbon of cytoplasm, in which vacuoles and pseudopodia containing PAS-positive granular material, arranged in the shape of a necklace, can be seen. This material is a neutrodiastase resistant mucopolysaccharide. These Sézary cells are considered giant histiocytes.

Syndromogenesis and etiology

This syndrome is considered to represent the leukemic phase of mycosis fungoides. Death occurs within 5 years consecutive to its appearance. Others think that although reticuloendothelial lesions are similar, this syndrome should be separated from mycosis fungoides.

The etiology is unknown.

Literature

Lutzner, M.A. and H.W. Jordan: The ultrastructure of an abnormal cell in Sézary's syndrome." Blood, 31:719-726, 1968.
Sézary, A. and Y. Bouvrain: Erythrodermie avec présence de cellules monstrueuses dans le derme et le sang circulant". Bull. French Soc. Derm. et Syph., 45:254-260, 1938.
Tedeschi, L.G. and D.T. Lansinger: «Sézary's Syndrome". Arch. Derm., 92:257-262, 1965.

SJÖGREN-LARSSON SYNDROME

Synonymy

S. of ichthyosiform erythroderma-spastic diplegia-oligophrenia.

Syndromography

Clinic

It does not occur preferentially in any sex, a history of consanguinity between relatives is recorded. It is characterized by presenting congenital ichthyosiform erythroderma. generalized but with little or no involvement of the scalp, hair or nails.

In addition, there is spastic cerebral diplegia typical of Little's disease, which is diagnosed from the first days of life; recurrent comeal ulceration, severe mental deficiency of the oligophrenia type (idiots or imbeciles), grand mal epilepsy, speech disorders, and degenerative retinitis.

Physical examination. Generally, they are detectable from birth: fine and normal hair, moderate hyperkeratosis of the skull and face, mild ichthyosis on the neck, back, trunk and limbs. Moderate hyperkeratosis on the palms

hands and soles of the feet. There may be dysplasia of the teeth and macular and corneal lesions in the eyes. From the neurological point of view, there is osteotendinous hyperreflexia especially in the legs, clonus of the patella, Babinski sign.

Paraclinical examinations

Pneumoencephalography. Hydrocephalus and cortical sclerosis with atrophy are observed. There are no calcifications.

Computed tomography and magnetic resonance imaging. The findings indicated in the pneumoencephalography are specified.

Skin biopsy. It allows checking hyperkeratosis and acanthosis, reduction of the stratum granulosum.

Syndromogenesis and etiology

It is an autosomal recessive syndrome. The cases described in Sweden are assumed to derive from a mutation dating back at least 600 years and the carrier frequency is 3%.

The basic defect is related to deficiency of the fatty aldehyde dehydrogenase enzyme, which catalyzes the oxidation of fatty alcohol to fatty aldehyde and fatty acid. Clinical improvement has been reported with restriction of fatty acids and supplementation of media with triglyceride chains.

Literature

Pardo-Castelló, V. and H. Faz: Ichthyosis-Little's disease". Arch. Derm., 26:915, 1932.

Rizzo, W.B. and D.A. Graft: "Sjögren-Larsson syndrome: deficiency te activity of the fatty aldehyde dehydrogenase component of fatty alcohol: NAD + Oxide reductase in culture fibroblasts". J. Clin. Invest., 88:1643-1648, 1991.

Romanes, G.J.: "Sjogren-Larsson syndrome." Brit. J. Ophthal., 52:174-177, 1968.

Sjögren, T. and T. Larsson: "Oligophrenia in combination with congenital ichthyosis and spastic disorders; clinical and genetic study." Psychiat Act. Neur. Scand., 32:Suppl. 113:1-112, 1957.

SNEDDON-WILKINSON SYNDROME

Synonymy

S. de Duhring-Sneddon-Wilkinson. S. of subcorneal pustular dermatosis.

Syndromography

Clinic

It predominates in women 4:1 and generally begins at age 40. It is rare in children. It is characterized by a pustular rash in the groin, armpits, and undermammary regions; flexor surfaces of the limbs and occasionally palms of the hands and soles of the feet. Sometimes it appears on the face and mucous membranes.
There is erythema around the pustule from the beginning, the pustules are generally oval, flaccid and cloudy in content, they dry out in a few days and leave a crusty surface.
Their grouping produces a serpiginous lesion.

Paraclinical examinations

Skin biopsy. The pustules are subcorneal and are covered by a thin layer of keratin; Its base is formed by granular tissue or pointed cells. In the fluid of the injury there are neutrophils, red blood cells and eosinophils. Light acanthosis.

Syndromogenesis and etiology

It may be a variant of Stevens-Johnson syndrome.

The etiology is unknown.

Literature

Sneddon, ILB. and D.S. Wilkinson: “Postular subcorneal dermatosis.” Brit. J. Derm., 68:385-394, 1956.

Sneddon, I.B. and D.S. Wilkinson: “Dermatose pustuleuse souscornée.” Bull. Soc. fr. Derm. Syph., 64:226-233, 1957.

SPANLANG-TAPPEINER SYNDROME

Synonymy

S. of alopecia-hyperhidrosis-tongue with corneal opacity.

Syndromography

Clinic

It occurs in both sexes between 5-20 years of age and is characterized by partial, total or localized alopecia; dystrophic nails; palmoplantar hyperkeratosis; normal teeth, but the tongue has corneal opacities.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Spanlang, H.: "Beitrage zur Klinik und Pathologie seltener Hornhauterkrankungen (Dystrophia adiposa corneal, Dystker atosis corneae congenita)". Ztschr. F. Augenheilk., 62:21-41, 1927.

SPIEGLER-FENDT SYNDROME

Synonymy

Spiegler-Fendt sarcoid. S. of Bäfverstedt. S. of miliary lymphocytoma. S. of benign lymphadenosis cutis. S. of lymphadenomatous

Syndromography

Clinic

It predominates in women 3:1 and can begin at any age, more frequently in the second to third decades.
It comes with two clinical varieties:

Variety I. Lymphocytoma cutis circumscriptum: there are no systemic symptoms, with the exception of some alterations in the urinary tract. Prominent red-purple or yellowish nodules appear located on the face (60%), preferably on the earlobes and wings of the nose. Less common in the scrotum, nipples and vagina. Regional lymph nodes are rarely enlarged and there is no splenomegaly.

Variety II. Eruption of several blue miliary nodules on the face, trunk and limbs. Itching especially in the summer months. There is no enlargement of the lymph nodes or splenomegaly.

Paraclinical examinations

Skin biopsy. It shows lymphocytic infiltration of the dermis, separated from the epidermis by normal connective tissue. Sometimes an accumulation of eosinophils is observed in the periphery of the lesions.

Syndromogenesis and etiology

It is considered a benign hyperplasia of the reticuloendothelial tissue of the skin, caused by trauma, insect bites, photosensitivity, which would act as triggering factors.

Literature

Fendt, H.: “Beitrage zur kenntniss der sogenannten sarcoiden Geswulsteder Haut.” Arch. f. Dermat. u Syph., 53:213-242, 1900.

Hofer, W.: "Benign lymphadenosis cutis." Arch. Klin, Exp. Derm., 203:23-40, 1956.

Spiegler, E.: "Ueber die sogenannte Starkomatosiscutis." Arch. f. Dermat. u Syph., 27:163-174, 1894.

STEVENS-JOHNSON SYNDROME

Synonymy

S. of pluriorificial erosive ectodermosis. S. of exudative erythema multiforme. S. of malignant bullous erythema. S. mucotaneocular.

Bullous erythema multiforme. Pluriorificial erosive ectodermosis. S. de Baader. S. de Fissinger-Rendu. S. de Neumann II. S. de Klauder.

Syndromography

Clinic

It occurs at any age, but is more common in the first and third decades and there is a slight predominance in the male sex. In 17% of cases, individuals had a history of good health and were not exposed to known exogenous factors. In 50% of the individuals had a history of vague symptoms of headache and upper respiratory symptoms or urinary tract infections and consumed some drug (see etiology).

Acute onset with fever of 39-40 °C, headache, discomfort and pain in the throat and mouth. Very soon the patients become worse and present a weak pulse, tachycardia, polypnea, prostration and joint pain.

This syndrome is characterized by vesicles that are preferentially located on the oral mucosa, tongue and lips. It is aggravated by the presence of pseudomembranes, blood loss, salivation and difficulty swallowing, which make feeding difficult. Bilateral conjunctivitis, corneal ulcers, epistaxis, rhinitis, and crusting of the nasal passages frequently develop.

The face, hands and feet are invaded by a hemorrhagic petechial or vesiculobullous eruption with inflammation of almost all natural orifices.

These blisters rupture and are surrounded by an inflammatory area with hemorrhagic scabs forming on the lips.

Physical examination. Vesiculobullous lesions on the skin accompanied by lesions in two or more mucous membranes (oral cavity, conjunctiva, genitourinary tract). Nikolski's sign is negative.

The skin lesion in the shape of an iris" or target" is pathognomonic.

It can last 1-2 weeks, or sometimes cause death.

Paraclinical examinations

Blood. Increase in erythrocyte sedimentation rate and leukocytes.

Chest radiology. Pneumonia is frequently found.

Biopsy. Hyperkeratosis, acanthosis, vesicles with eosinophils, fibrin precipitate, lymphocytes and neutrophils. Perivascular lymphocytic infiltrate in the dermis.

Syndromogenesis and etiology

There are doubts whether it is due to one or multiple etiological factors (the most accepted): infectious, toxic, parasitic, fungal, drugs, etc.

In some cases there is an unequivocal correlation with drug administration. The most common are sulfas and penicillin, although some sedatives have also been reported.

Viral infections and some allergic mechanisms have been considered as etiological factors.

It seems to be a sensitization in the small cutaneous blood vessels, for any of the varieties of allergens or toxic substances.

The etiology remains obscure or unknown.

Literature

Bianchine, JR; P.V.J. Macaraeg; L. Lasagna et al: "Drugs as etiologic factors in the Stevens-Johnson syndrome." Amer. J. Med., 44:390-405, 1968.

Stevens, A.M. and F.C. Johnson: "A new eruptive fever associated with stomatitis and ophthalmia. Report of two cases in children." Amer. J. Dis. Child, 24:526-533, 1922.

STRYKER-HALBEISEN SYNDROME

Synonymy

S. of erythroderma-macrocytic anemia.

Syndromography

Clínic

It is characterized by weakness, fatigue and intense itching accompanied by skin lesions in the form of scaly patches and vesicular erythroderma, located on the face, neck and upper chest.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Stryker, G.V. and W.A. Halbeisen: "Determination of macrocytic anemia as an aid in diagnosis of certain deficiency dermatoses." Arch. Dermat. Syph., 51:116-123, 1945.

SULZBERGER-GARBE SYNDROME

Synonymy

S. of polymorphous prurigo. Sulzberger-Garbe chronic lichenoid dermatosis. S. of exudative lichenoid discoid dermatitis. Oid-Oid disease.

Syndromography

Clinic

It predominates in men, especially Jews, and begins abruptly between the fourth and sixth decades. They are generally patients with cyclothymic or neurotic personality.

It presents with intense nocturnal or paroxysmal itching located in the genitals, under the breasts, around the mouth or on the limbs; It often begins after a local infection.

Physical examination. Ephemeral exudative lesions, follicular papules and urticaria-like lesions. Lichenified and crusted plaques and other alterations may develop. The authors describe 4 evolutionary phases in this syndrome: 1, exudative and discoid plaques; 2, lichenified phase or papules; 3, prefungal phase; 4, urticarial phase.

Paraclinical examinations

Skin biopsy. The lesions are similar to seborrheic dermatitis with crusted lesions and lichenification.

Syndromogenesis and etiology

It is considered that there may be emotional factors that favor skin reactions and give rise to lesions:
scabby, reddish, etc.

Infectious eczematoid dermatitis has been described within this syndrome. It is considered an example of skin self-sensitization to different bacterial or chemical agents.

The etiology is unknown.

Literature

Sulzberger, M.B. and W. Garbe: "Nine cases of a distinctive exudative discoid and lichenoid chronic dermatosis." Arch. Derm. Syph.., 36:247-278, 1937.

SWEET SYNDROME

Synonymy

S. of acute febrile neutrophilic dermatosis.

Syndromography

Clinic

It has only been reported in adult women and is characterized by high, persistent fever and the appearance of raised, painful erythematous plaques on the skin of the limbs, face and neck.
Pustules subsequently form.

Paraclinical examinations

Blood. Leukocytosis with marked neotrophilia.

Skin biopsy. Dermal infiltration is observed, consisting of mature polymorphonuclear neutrophils. Moderate vasodilation and edema of the vascular endothelium.

Syndromogenesis and etiology

This is possibly a hypersensitivity reaction. No infection has been demonstrated, but he responds quickly to steroids.

Literature

Sweet, R.D.: "An acute febrile neutrophilic dermatosis." Br.J. Derm., 76:349-356, 1964.

BLACK HEEL SYNDROME

Syndromography

Clinic

Predominates in female adolescents. It generally appears in epidemic form. It is characterized by a blackish discoloration of isolated or associated plaques on the heels or adjacent regions. Rarely in the metatarsal region.

Syndromogenesis and etiology

It could occur due to blood extravasation. There is no explanation why it occurs in epidemic form.

The etiology is unknown.

Literature

Bazex, A.; R. Sal vador and A. Dupre: "Plantar chromhidrosis." Bull. French Soc. Derm. Syph., 69:489-490, 1962.

THEVENARD SYNDROME

Synonymy

Familial S. ulceration of the limbs. S. of familial mutilating ulcerative acropathy. S. of neuropathic acrodystrophy. Bad plantar perforation.

Syndromography

Clinic

It begins at puberty and is characterized by:

1. Trophic ulcers of the soles of the feet.
2. Hypoesthesia to pain and temperature in the soles of the feet.
3. Osteodystrophies of the feet (elephant feet) and spine.
4. Vasomotor disorders.
5. Hypertrichosis

Paraclinical examinations

Radiology. Injuries are limited to limbs lower.

Syndromogenesis and etiology

It has not been possible to identify a specific neurological lesion. It is transmitted by autosomal dominant inheritance.

The etiology is apparently genetic according to the inheritance pattern indicated.

Literature

Banna, M. and J.B. Foster: "Roentgenologic features of acrodystrophic neuropathy." Am. J. Roentgenol., 115:186, 1972.
Thévenard, A.: Lácropathie ulcero-mutilante familiale". Rev. Neurol. (Paris), 74:193, 1942.

TIETZ SYNDROME

Syndromography

Clinic

It is characterized by:

1 Albinism.

2. Complete deafness.

3. Hypoplasia of the eyelashes and eyebrows, with normal eyes.

Syndromogenesis and etiology

The etiology is unknown.

TYLOSIS-OPTICAL ATROPHY SYNDROME

Syndromography

Clinic

It is convenient to clarify that the term tylosis is used synonymously with keratoderma, keratoma, hyperkeratosis.

Predominates in women. It is characterized by palmoplantar keratoderma and occasionally in the ears. Late in life, patients develop optic atrophy.

Syndromogenesis and etiology

It is transmitted by intermediate inheritance linked to sex.

It is good to note that recessive forms of palmoplantar tylosis or keratosis are generally part of other ectodermal anomalies and give rise to different syndromes. (See syndromes associated with tylosis.)

The etiology is unknown.

Literature

Dimsdale, H.: "Hereditary optic atrophy in families with keratoderma palmaris et plantaris (tylosis)." Proc. Royal Soc. Med., 42:796, 1949.

TYLOSIS SYNDROME -ESOPHAGEAL CARCINOMA

Syndromography

Clínica

It occurs in both sexes. It is characterized by palmoplantar keratosis that begins between the third and sixth decades and during its evolution, an esophageal carcinoma is detected, generally located in the lower third. (Howel-Evans has described six cases in the same family.)

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Howel-Evans, W.; R.B. McConnell; AC Clarke et al.: Carcinoma of the esophagus with keratosis palmaris et plantaris (tylosis). A study of two families". Quart. J. Med., 27:413-429, 1958.

Shine, I. and P.R. Allison: "Carcinoma of the esophagus with tylosis (keratosis palmaris et plantaris)." Lancet, 1:951-953, 1966.

TOURAINE-SOLENTE-GOLÉ SYNDROME

Synonymy

S. de Brugsch. S. of acropachydermia. S. of pachydermoperiostosis. Osteodermopathic S. S. of osteodermopathy megalia cutis et osseum. (See dysacromelias and tuberculosis.)

Syndromography

Clinic

This syndrome is almost exclusive to men and begins after puberty in the third decade. The skin on the forehead, face, skull, hands and feet begins to become thick and wrinkled. There is hyperhidrosis of the hands and feet with increased sebaceous secretion. Hands and feet grow intensely. The nails are like watch glass and the fingers are drumsticks.

The arms and legs are cylindrical and there are effusions in the knees and ankles or other joints. The general condition and mental health are not affected.

Paraclinical examinations

Bone radiology. It allows us to specify the magnitude of the growth of the hands and feet. Skeletal and chest radiographs are also useful, showing peripheral periostitis with diffuse and irregular periosteal ossification.

Skin biopsy. Hypertrophy of the connective tissue and epidermis.

Syndromogenesis and etiology

The primary form is transmitted by autosomal dominant inheritance with variable expression related to sex. The secondary form, characterized by hypertrophic osteoarthropathy, is similar or identical to Marie-Blumberg syndrome (see).

Three forms of presentation are recognized: complete form (periostosis and pachyderma), incomplete form (without pachyderma) and frustrated form (pachyderma with minimal skeletal changes).

Genetic heterogeneity is suggested.

Literature

Brugsch, H.G.: "Acropachyderma with pachyperiostitis; report of a case." Arch. Int. Med., 68:687-700, 1941.

Labbe, M. and P.Renault: “Hypertrophic osteodermapathy.” Bull. et Mem. Soc. Méd. d. Hop. de Paris, 1:1065-1067, 1926.

Touraine, A.; G. Solente and L. Golé: *Un syndrome osteodermapathique: la pachyperiostose des extremités". Presse Méd., 42:1820-1824, 1935.

CONGENITAL TRICOMEGALY SYNDROME

Syndromography

Clinic

It commonly develops in the first months of life.

It is characterized by bilateral destruction of the tear ducts. There is slow dentition, poor vision with pigmentary degeneration of the retina, hypertrichosis of eyelashes and eyebrows, alopecia between 4-5 years of life, dwarfism, mental retardation or normality.

Syndromogenesis and etiology

It is considered a form of ectodermal dysplasia. There is no family history.

The etiology is unknown.

Literature

Cant, J.S.: "Ectodermal dysplasia." J. Pediat. Ophthal., 4:13-17, 1967.

Oliver, G.L. and D.C. McFarlane: "Congenital trichomegaly with associated pigmentary degeneration of the retina, dwarfism, and mental retardation." Arch. Ophthal., 74:169-171, 1965.

YELLOW NAIL SYNDROME

Syndromography

Clinic

It is characterized by yellowish discoloration of the nails, which are also slow growing and show loss of the lunula and cuticle. This syndrome is accompanied by paronychia, onycholysis and malleolar edema.

Sometimes there may be edema of the face and hands, pleural effusion and hypoalbuminemia. Sometimes yellow nails can be associated with syphilis.

Syndromogenesis and etiology

External exposure to resorcin, anthralin or chrysarrobin can induce the appearance of yellow nails.

The etiology is unknown.

Literature

Heller, J. in Jadassohn: Handbuck der Hautund Geschlechtskranhheiten XII, Berlin, Springer, 1927.

Parks, R. and J.P. Ellis: *Yellow Nails", Arch Derm., 102:619-623. 1970.

Samman, P.D. and W.E White: "The ´yellow nails´ syndrome". Brit. J. Derm., 76: 153-157, 1964.

PLINER NAIL SYNDROME

Syndromography

Clínic

It is characterized by intense pain in the affected fingers.

Upon examination, an excessive transverse curvature is observed in the nails of these fingers with loss of their soft tissue.

Paraclinical examinations

Radiology. Examination of the fingers shows bone resorption in the affected phalanges.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Cornelius, C.E. and W.B. Shelley: “Pincer nails syndrome.” Arch. Surg., 96:321-322, 1968.

Samman, P.S.: The Nails in Disease. London, Heiremann, 1965, p. 79.

URBACH-WIETHE SYNDROME

Synonymy

S. of hyalinosis cutis et mucosae. S. of lipoid proteinosis. S. of lipoglycoproteinosis. S. de Rössle-Urbach-Wiethe.

Syndromography

Clinic

It can be congenital, but it generally begins around age 6; However, most cases occur in the elderly. It is characterized by small yellowish-white, waxy papules, in the form of nodules and/or plaques that can become infected and ulcerate, and leave varicose-like scars. They are located on the face, neck, armpits, hands, eyelid margins, scrotum. They are accompanied by keratosis on the elbows and knees.

There may be ivory-colored spots such as "morphea", alopecia and gray hair.

They also affect the mucous membranes, especially the mouth (lower lip), larynx (vocal cords), esophagus and vagina.

They can cause dysphonia and dysphagia. When the nodules occlude the Stenon's duct, painful swellings of the parotid glands occur. After several years, brain calcifications may occur, causing epileptic seizures.

Paraclinical examinations

Skin biopsy. Hyalinosis of the upper layers of the dermis that begins perivascularly is observed.

Blood. Hypercholesterolemia and increased betalipoproteins.

Radiology. Cranial calcifications (25%) bilaterally in the hippocampus, falx cerebri and temporal lobes.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance. It is considered to be due to a degeneration of collagen and elastic fibers that are infiltrated by lipids or galactolipids.

The etiology is unknown.

Bibliography

Burnett, J.W. and S.M. Marcy: "Lipoid proteinosis." Am. J. Dis. Child., 105:81, 1963.

Gordon, H. et al.: Lipoid proteinosis." Birth Defects, 7(8): 164, 1972.

McCusker, J.J. and R.M. Caplan: "Lipoid proteinosis (lipo-glycoproteinosis): a histochemical study of two cases." Amer. J. Path., 40:599-613, 1962.

Urbach, E. and C. Wiethe: Lipidosis cutis et mucosae", Virchows Arch. Path. Anat., 273: 285-319, 1929.

URTICARIA PIGMENTOSA SYNDROME

Synonymy

S. of mastocytosis. S. of Nettleship.

Syndromography

Clínic

In half of the cases, it generally begins from the first months of birth; 25% before puberty.

It is characterized by skin lesions that may consist of:

macules, papules, nodules, warts and vesicles.

Telangiectasias, petechiae or ecchymoses are rarely present. At their beginning, the lesions are similar to urticaria, but they are not evanescent like them; On the contrary, they persist and slowly acquire a slate color.

The most frequent location of these lesions is on the limbs and trunk and they almost always spare the face. If they are rubbed intensely they can cause a raised lesion. Bruising or excessive scratching sometimes causes serious symptoms as a result of the release of histamine. Itching is the most constant symptom and can vary from mild to intense, which interferes with sleep. Old lesions tend to disappear over time without leaving any sequelae, but the rash remains and new ones develop, although there are periods free of the condition. Two clinical forms have been observed: juvenile and adult, including some clinical varieties.

Paraclinical examinations

Skin biopsy. It shows in the dermis a dense accumulation of large mononuclear cells with abundant basophilic cytoplasm containing metachromatic granules.

The floor of the blister is sub-epidermal and the mast cells or mast cells are grouped in a strip below and to the sides of the vesicle, but on the outside of the skin.

Syndromogenesis and etiology

Autosomal dominant inheritance seems to be the most likely transmission.

In this syndrome, mast cell infiltrates are found in the superficial and middle layers of the dermis, particularly around the vessels. Sometimes also visceral mastocytosis: the release of histamine from these cells explains much of the symptoms.

An abnormal ultrastructure of mast cell granules has been reported when observed with the electron microscope.

Literature

Anstey, A.; D.G. Lowe and J.D. Kirby: M.A. Horton: Familial mastocytosis: a clinical, immunophenotypic, light and electron microscopic study." Br. J. Derm. 125:583-587.

Burgoon, C.F. Jr.: JH. Graham and D.L. Mc Caffre: "Mastcell disease. "A cutaneous variant with multisystem involvement".
Arch. Dermatol., 98:590, 1968.

Nettleship, E.: "Rare forms of urticaria." Br. Med. J., 2: 323, 1869.

Perlman, H.R.: Pediatric Dermatology. Ed. Year Book, Chicago, 1960. p. 139.

Selmanowitz, V.J. and N. Orentreich: “Mastocytosis a clinical genetic evaluation.” J. Hered., 61:91, 1970.

NODULAR VASCULITIS SYNDROME

Syndromography

Clinic

It predominates in women between 30-50 years old (without evidence of tuberculous disease). It is characterized by chronic painful nodules of red color and varying in size between a few millimeters and centimeters in diameter, isolated or in clusters that are located on the limbs, generally on the legs below the knees; Sometimes they are arranged linearly. The nodules may or may not ulcerate and recurrences are common, especially when patients return to work.

There is frequently a history of long-term phlebitis. Eventually brown pigmentation may develop along the superficial veins.

Paraclinical examinations

Skin biopsy. It rules out a tuberculous lesion and shows vasculitis with different degrees of obliteration of arterioles and venules, necrosis of the subcutaneous fat and fibrosis. When a large artery or vein is the center of an infiltrative reaction, the diagnosis of nodular vasculitis is correct.

Syndromogenesis and etiology

It was previously considered a form of Bazin syndrome.

Currently, as a vasculitis secondary to bacterial infections (Parishy Rhodes).

The etiology is unknown.

Literature

Audry, C.: "Etude de la lesion de lérytheme indure (de Bazin) sur la notion du lymphatisme." Ann. from Dermat. et Syph., 29:209-214, 1898.

Montgomery, H.; P.A. O'Leary and N.W. Barker: "Nodular vascular diseases of the legs: Erythema induratum and allied conditions."
J.A.M.A., 128:335-341, 1945.

Montgomnery, H.: Dermatopathology. New York, Hoeber, 1967.

VILANOVA-AGUADÉ SYNDROME

Synonymy

S. of migratory nodular panniculitis. S. of sclerodermiform panniculitis.

Syndromography

Clinic

This syndrome is rare in men and frequently presents a history of acute tonsillitis. It can also be preceded by trauma.

It is characterized by nodules or plaques, single or grouped, that appear on a leg, generally in the malleolar region, and expand to the joints forming a long erythematous plaque that becomes hard with successive eruptions. It lasts for months or years, always limited to the legs.

Paraclinical examinations

Blood. Elevated erythrocyte sedimentation rate. Positive rheumatoid factor and increased antistreptolysin titer.

Skin biopsy. Epithelial proliferation that occludes capillaries and small vessels. Infiltration with lymphocytes, monohistiocytes and fibroblasts. Later, only giant cells are observed. The lesions disappear with Restitutio ad integrumn when they heal.

Syndromogenesis and etiology

It is thought that it may be a variety of nodular vasculitis.

The etiology is unknown.

Literature

Vilanova, X. and J.P. Piñol Aguadé: "Hypodermite nodulaire subaigué migratrice." Ann. Derm. Syph., 83:369-404, 1956.

Vilanova, X. and J.P. Piñol Aguadé: "Subacute nodular migratory panniculitis." Br. J. Derm., 71:45-50, 1959.

VOHWINKEL SYNDROME

Synonymy

S. of keratoderma mutilans.

Syndromography

Clínica

It occurs in both sexes and is characterized by hyperhidrosis, keratosis pilaris, watch-glass nails, associated with the development of tylosis, and fissures with constrictions reminiscent of Ainhum syndrome (see this syndrome), which lead to spontaneous amputation of the fingers. There are also symptoms and signs of hypogonadism. Years after the onset of tylosis, symptoms and signs of syringomyelia develop. Hearing loss of cochlea origin.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. The basic defect is not known. Histological examination of the skin biopsy has shown marked orthohyperkeratosis with thickening of the basal and granular laminae.

The etiology is genetic.

Literature

Sensi, A.; V. Bettoh et al.: Vohwinkel syndrome (Mutilating Keratoderma) associated with craniofacial anomalies". Am. J. Med. Genet., 50:201-203, 1994.

Tatz, K.: “Pityriasis rubra pilaris with ainhum and syringomyelia.” Br. J. Dermat., 58:123-126, 1946.

Vohwinkel, K.H.: "Keratoma hereditarium mutilans." Arch. Derm. u Syph., 158:354-364, 1929.

WEBER-CHRISTIAN SYNDROME

Synonymy

S. de Pfeifer-Weber-Christian. Recurrent non-suppurative febrile nodular panniculitis. Weber disease- -Christian.

Syndromography

Clinic

It occurs in both sexes and can appear at any age. It is preceded by prodromal symptoms: general malaise, low-grade fever. In addition, there is oropharyngeal infection, mild arthralgia or arthritis.

The typical clinical picture is given by recurrent attacks of fever and general malaise accompanied by subcutaneous nodules of an inflammatory nature, mobile and painful, mainly distributed on the limbs and trunk. Subcutaneous atrophy is also observed at the sites of the nodules when the acute condition subsides.

Physical examination. Subcutaneous nodules of different diameters (between 1-12 cm) are seen anywhere on the body, most frequently on the thighs, hands, face, and feet. They are usually isolated. They are sensitive but not painful. The skin in the nodules takes on a reddish color. After the acute phase, the skin appears pigmented and atrophies. The nodules rarely rupture and release a yellow greasy fluid. In some cases there is splenomegaly. The lesions recur frequently.

It can simultaneously affect the ligaments, mesentery and perivisceral fat, and is associated with pancreatitis, pancreatic carcinoma and other diseases of this or other organs.

Paraclinical examinations

Skin biopsy. At first it shows macrophages with fatty inclusions. Later, small areas of central necrosis or fat necrosis surrounded by polymorphonuclear lymphocytes surrounded by macrophages. Over time, the necrotic material decreases and fibrotic changes appear.

Syndromogenesis and etiology

The etiology of this syndrome is unknown. The following possibilities have been raised:

- Drug allergy.
  Reticuloendothelial response to bacteria or their toxins.
  Viral infections.
  Vascular alterations.
  Collagen alteration.
  Trauma.
- Hypersensitivity to iodine and bromides.
- Septic sources.
  Autoimmune processes.

Literature

Christian, H.A.: “Relapsing febrile, nodular nonsuppurative panniculitis.” Arch. Int. Med., 42:338-35 1, 1928.

Leonhardt, T.: “A case of Weber-Christian disease roentgenographically demonstrable mammary calcifications.” Amer. J. Med.,44:140-146, 1968.

Pfeifer, V.: Uber einen Fall von herweiser Atrophie des subkutanen Fettgewebes". Deutsch. Arch. Klin. Med., 50:438-449, 1892.

Popoff, N. and M. Wheelock: “Relapsing febrile nodular nonsuppurative panniculitis; Pfeifer-Weber-Christian disease; report of 3 cases". A.M.A. Arch. Int. Med., 97:39, 1956.

Weber, FP.: “A case of relapsing nonsuppurative nodular panniculitis showing phagocytosis of subcutaneous fat cells by macrophages.” Br. J. Derm., 37:301-3 11, 1925.

WEBER-COCKAINE SYNDROME

Synonymy

S. of localized epidermolysis bullosa.

Syndromography

Clínic

The blistering eruptions in this syndrome are recurrent and are located on the hands and feet. They are almost always familial and appear chronically, during infancy and later. Usually, similar cases are recorded in the family. The injuries are exacerbated by hot weather and when the patient is subjected to prolonged marches, as occurs in military service. It is important to note that injuries do not leave scars.

Occasionally it is associated with hyperhidrosis. Injuries can be prevented by applying ice to friction areas.

Syndromogenesis and etiology

This syndrome of genetic etiology constitutes an example in man of a heat-sensitive mutation, with autosomal dominant transmission.

Linkage has been found on chromosome 12q and 17q close to the keratin gene region.

Literature

Bonifas, J.M.; TO THE. Rothmann; E. Epstein: “Linkage of epidermolysis bullosa simplex to probes in the region of eratin gene cluster on chromosomes 12q and 17q.” (Abstract). Clin. Res., 39: 503A. 1991.

Johnson, S. and A. Test: Epidermolysis bullosa simplex of the hands and the feet. A genetic study of the Hereditary type". Arch. Dermat. & Syph., 53:610, 1946.

WEIR-MITCHEL I SYNDROME

Synonymy

S. de Gerhardi II. S. of erythromelalgia. S. of erythelmalgia.

Syndromnography

Clinic

It is characterized by a burning sensation in the limbs, with redness and an increase in temperature, often coinciding with an increase in ambient temperature, especially in the summer. It is more common at night when the patient is lying down. Disappears with cooling. Trophic changes occur.

Paraclinical examinations

Blood. It is necessary to perform tests to rule out or confirm myeloproliferative syndromes.

Heat test. Clinical symptoms are reproduced.

Syndromogenesis and etiology

This syndrome frequently precedes the onset of a myeloproliferative syndrome by years.

The etiology is unknown.

Literature

Alarcón-Segovia, D.; R.R. Babb: J.F. Fairbairn II et al.: "Erythermalgia. A clue to the carly diagnosis of myeloproliferative disorders". Arch. Int. Med., 117:511-5 15, 1966.

Mitchell, S.W.: "Clinical lecture on certain painful affections of the feet." Philadelphia, M. Times, 3:81-82, 1872.

WHITFIELD SYNDROME

Syndromography

Clinic

It predominates in the female sex and begins in middle age. It is characterized by a feeling of restlessness or discomfort in the legs, after having been standing for a long time, accompanied by general discomfort.

Physical examination. It shows edema of the ankles, tender nodules with painful stabs after exposure to cold, trauma or general infections. No erythrocyanosis or ulcerations are observed.

Paraclinical examinations

Skin biopsy. It shows different degrees of vasculitis with seizure of the venules; edema and fibrous changes.

Syndromogenesis and etiology

It is hypothesized that venous stasis may play a role; Allergic factors have also been considered. (See the syndromes of: Bazín and Vilanova-Aguadé.) The etiology is unknown.

Bibliography

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

Whitfield, A.: *On the nature of the disease known as erythema induratunm seroful osorum". Amer. J. Med. Sci. n.s., 122:828-834, 1901.

WILLIAM-PLUMBE SYNDROME

Synonymy

Psoriatic S.

Syndromography

Clínic

It occurs in both sexXOs and at any age. It is rare before the age of 3, it increases during puberty. Typical lesions appear after an infection or trauma in any area of the skin, usually on the face.

Physical examination. Uniform, thick, red lesions, of different morphology, exfoliative, pustular, elevated.

They can affect the nails or subungual regions and are sometimes associated with arthritis.

Two clinical syndromes have been recognized:

1 Erythrodermic psoriatic syndrome, the exfoliative phase appears suddenly, or evolves chronically.
2. Unstable erythrodermic psoriatic syndrome, appears after an infection.

Paraclinical examinations

Blood. Hypocalcemia and/or hyperuricemia in some cases.

Skin biopsy. Micropustules, acanthosis, hyperkeratosis, polymorphonuclear invasion.

Syndromogenesis and etiology

Familial predisposition, autosomal recessive inheritance.

The etiology is unknown.

Literature

Plumbe. S.: Practical Treatise of the Skin. London, Underwood, 1824.

Rook, A.; D.S. Wilkinson and F.J.G. Ebling: Textbook of Dermatology. Philadelphia, Davis, 1968.

William, R.: Description and treatment of cutaneous diseases. London, 1:132-188, 1808.

WITKOP-VON SALLMANN SYNDROME

Synonymy

S. hereditary benign intraepithelial dyskeratosis.

Syndromnography

Clinic

It begins in the infant with ocular lesions in the form of gelatinous and spongy plaques in the ocular conjunctiva, which

They can lead to blindness by invading the cornea. Features:
mouth corners covered with white plaques. Mouth: similar plaques on the lips, gums, lateral and ventral surfaces of the tongue, floor of the mouth, palate and fauces, with the appearance of leukoplakias.
There does not seem to be a tendency toward malignancy in these areas.

They generally progress until adolescence, and then stabilize.

Paraclinical examinations

Skin biopsy. Characteristic histological changes of the sheet of spiny cells of the mucosa, which presents numerous curious-looking areas of eosinophilic cells that appear to be engulfed by normal cells, giving the appearance of cells within cells.

Syndromogenesis and etiology

It is transmitted by dominant inheritance with great penetrance that reaches 97%.

The etiology is monogenic genetics.

Literature

Goodman, R.M. and R.J. Gorlin: Atlas of the Face in Genetic Disorders. Ed. Mosby, St. Louis, 1977. p. 130.

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Von Sallmann, L. and D. Paton: "Hereditary benign intraepithelial dyskeratosis: Ocular manifestations." Arch. Opthal., 84:762, 1961.

Witkop, C.J. Jr.; C.H. Shankle; J.B. Graham et al.: "Hereditary benign intraepithelial dyskeratosis." Arch. Path., 70:696, 1960.

ZOON SYNDROME

Synonymy

S. of balanitis due to plasma cells.

Syndromography

Clinic

It is observed in middle-aged or old men. It is characterized by the presence of painless plaques on the foreskin and glans, moist, shiny, with dots on the skin in that area that simulate peppercorns.

Paraclinical examinations

Skin biopsy. Infiltration of plasma cells and hemosiderin deposits.

Syndromogenesis and etiology

It is possible that it is caused by chronic nonspecific balanitis.

The etiology is unknown.

Literature

Zoon, J.J.: "Balantis circumscripta chronica met plasmacellen-infiltrant", Ned. Tschr. Geneesk., 94:1529-1530, 1950.

RELATED DERMATOLOGICAL SYNDROMES

Syndromes with alopecia-ipotrichosis

Hydrotic ectodermal dysplasia.
Hallermann-Streiff.
Cartilage-hair hypoplasia.
Hutchinson-Gilford.
Laurence-Moon-Biedl.
Marinesco-Sjögren.
Netherton.
Rothmund.
Wedderburn.
Werner.

Syndromes with blisters or blisters

Senile herpetiform dermatitis.
Duhring-Brocq.
Hereditary fetal epidermolysis bullosa.
Hyperplastic epidermolysis bullosa.
Epidermolysis bullosa polydystrophy.
Epidermolysis bullosa simplex.
Hallopeau.
Lortat-Jacob-Degos.
Lyell.
Neumann.
Pasini.
Juvenile pemphigoid.
Pemphigus vulgaris.
Senear-Usher.
Sneddon-Wilkinson.
Stevens-Johnson.

Dwarfism syndromes-dermatological manifestations

Blom.
Cokayne.
Bird-headed dwarfism (Seckel syndrome).
Hallermann-Streiff.
Hutchinson-Gilford.
Rothmund-Thompson.
Werner.

Syndromes with sclerodermia

Buschke.
Circumscribed calcinosis.
Universal calcinosis:
CRST.
Dermatomyositis (changes may be observed sclerodermic).
Systemic sclerosis.
White spots.
Morphea.
Lupus erythematosus.

Syndromes with anhidrotic ctodermal dysplasia

Cockayne.
Berlin.
Goltz-Gorlin.
Hellweg-Larsen.
Ito.
Marshall.
Naegeli I.
Wedderburn.

Syndromes with hyperhidrotic ectodermal dysplasia

Böök
Brunaue.

Fisher.

Jadassohn-Lewandowsky.

Meleda.

Papillon-Lefevre.

Schwann.

Siemens II.

Spanlang-Tappeiner.

Vohwinkel.

Zinsser-Engman-Cole.

Syndromes with normohidrotic ectodermal dysplasia

Bloch-Sulzberger.

Capdepont.

Cranco-ectodermal dysplasia.

Ellis-van Creveld.

Gorlin-Cohen.

Gorlin-Chaudhry-Moss.

Grob.

Hallermann.

Hutchinson-Gilford.

Oculodentdigital.

Rothmund.

Thompson.

Unna-Thost.

Van Bogaert-Hozay.

Werner.

Syndromes with hypermelanosis

Hereditary:

Albright.

Bloch-Sulzherger.

Fanconi.

Gaucher.

Mendes Da Costa.

Naegeli.

Peutz-Jeghers.

From Sanctis-Cacchieone.

Von Recklinghausen I.

Non-Hereditary:

Addisonian.

Basedow.

Cushing.

Dermatomyositis.

Hanot-Chauffard.

Hodgkin.

Malnutrition.

Marie.

Schilder.

Syndromes with idiopathic lipogranulomatous hypodermitis

Bazin.

Darier-Roussy.

Periarteritis nodosa of the skin.

Rothmann-Makal.

Weber-Christian.

Syndromes with ichthyosis

Ichthyosiform erythroderma.

Lamellar ichthyosis.

Hysterical ichthyosis.

Dominant vulgar ichthyosis.

Sex-linked ichthyosis vulgaris.

Bullous-ichthyosiform hyperkeratosis.

Mendes da Costa.

Collodion child.

Netherton.

Refsum.

Rud.

Sjögren-Larsson.

Neurocutaneous syndromes

Autonomous clinical forms:

Bonnet-Dechaume-Blanc.

Bourneville.

From Sanctis-Cacchione.

Leschke.

Louis-Bar.

Pringle.

Recklinghausen.

Schimmelpenning-Feuerstein-Mins.

Sturge-Weber.

Von Hipple-Lindau.

Other Syndromes:

Albright.

Bean.

Bloch-Sulzberger.

Brooke.

Capute-Rimoin-Konigsmark.

CurtiusI.

Gingival fibromatosis-hypertrichosis.

Flinn-Aird.

Gorlin-Goltz.

Godfried-Prick-Carol-Prakken.

Grönblad-Strandberg.

Ito.

Klippel-Trenaunay.

LEOPARD.

Mafucci.

Neurocutaneous melanoblastosis.

MMN.

Murray.

Naegeli I.

Oldfield.

Osler I.

Ota.

Familial polyposis of the colon.

Juvenile polyposis.

Turcot.

Van Bogaert-Divry.

Zanca.

Unusual odor syndromes

Isovaleric acidemia.

Folling.

Hypermethioninemia (fish, sweets, fruits, butter

lla stale roucid butter, boiled cabbage).

Smell: cheese, sweaty feet.

Oast-house.

Smell: dried maltor hops.

Smell: musty wolf-like, barry, stale sweaty locker-room towel.

Smell: caramel, malt, malt syrup.

Maple syrup.

Syndromes with panniculitis

Dercum.

Cold panniculitis in children.

Rothmann-Makal.

Vilanova-Aguadé.

Weber-Christian.

Syndromes with poikiloderma

Symptomatic poikiloderma as the final phase of dermatosis.

Actinic dermatosis.

Dermatomyositis (Petges-Cléjat, Wagner-Unverricht).

Scleroderma

Mycosis fungoides (Alibert-Bazin).

Vulgaris psoriasis.

Pityriasis rosea.

Lupus erythematosus.

Bloom.

Idiopathic poikiloderma:

Riehl.

Congenital poikiloderma:

Rothmund.

Congenital ectodermal dysplasia with cataracts (Cole, Giffen, Simmons and Stroud).

Thomson.

Zinsser-Engman-Coley Fanconi-Zinsser.

Marghescu-Braun and Falco-Rodermund.

parotid

Greither.

Van Bogaert-Divry.

Dubowitz.

Cockayne.

Gottron I.

General:

Werner.

Buschke-Ollendorff.

Papillon-Léage-Psaume.

Christ-Siemens-Touraine.

Metagery.

From Sanctis-Cacchione.

Louis-Bard.

Syndromes with keratoses

Meleda.

Papillon-Lefebre.

Vohwinnkel.

Nockemann.

Siemens.

Fisher.

Brünauer.

Spanlang-Tappeiner.

Richner-Hanhart.

Volavsek.

Schäfer.

Hanhart IV.

Naegeli I.

Bloch-Sulzberger.

Unna-Thost.

Jadassohn-Lewandoswky.

Zinsser-Engman-Colf.

Kumer-Loos.

Bureau.

Haxthausen.

Böök.

Capdepont.

Bauer.

Greither.

Gorlin-Goltz.

Schwann.

Hellweg-Larsen

Darier I.

Rothmund.

Werner.

Thomson.

Christ-Siemens-Touraine.

Berlin.

Clarke.

Ichthyosis vulgaris.

Acanthosis.

Siemens I.

Marrow-Brooke.

Congenital ichthyosiform erythroderma.

Riehl.

Gonorrheal keratosis.

Kogoj.

Ainhum.

Kyrle.

Syndromes with tylosis

Ainhum.

Pads on the knuckles-deafness-leukonychia.

Berlin.

Brauer.

Brunauer.

Dentinogenesis imperfecta.

Ectodermal hydroic dysplasia.

Fisher-Volasek.

Franceschetti-Thier.

Greither.

Helweg-Larsen.

Jadassohn-Lewandosky.

Meleda.

Nayeli.

Papillon-Lefevre.

Climacteric keratoderma.

Schafer.

Spanlang-Tappeiner.

Tylosis-carcinoma of the esophagus.

Tylosis optic atrophy.

Vohwinkel.

Wedderburn.

PERIODIC ABDOMINALGIA SYNDROME

Synonymy

S. of Siegal-Cattan-Mamou. Periodic peritonitis. S. of benign paroxysmal peritonitis. Recurrent familial polyserositis. Familial Mediterranean fever. S. de Wyllie-Schlesinger.
Reismann's periodic disease.

Syndromography

Clínic

Peculiar distribution in people with ancestors of the Mediterranean

About 60% of affected patients are men.

Symptoms begin between 5 and 15 years of age. The typical acute crisis lasts 24-48 hours, although it can last for a week. It can occur with an interval of one crisis every 2-4 weeks.

Late onset in adolescence, sometimes earlier with more intensity.

Abdominal pain of colicky type, of variable intensity and location. Accompanied by fever, sometimes cyclical vomiting, paleness, headache.

Fever is a common symptom and appears in almost all attacks. It is preceded by episodes of chills with fever ranging from 38.5-40 °C.

Abdominal pain is common, appears in one quadrant and then radiates to the entire abdomen. It can be associated with pain in both shoulders that expresses diaphragmatic irritation. Nausea and vomiting.

Arthralgia lasting 3-5 days in large joints in 60-75% of patients.

Physical examination. Pain, diffuse or not, in the abdomen and signs of moderate distension.

There is pain on palpation, contracture and decreased air fluid sounds. Positive decompression maneuver.

Chest: Unilateral stabbing pain and dyspnea in 40-75% of patients.

cases.

Decrease in vesicular murmur. Episodes of unilateral pleuritis, pleural friction rub and transient effusion.

Joint pain, with acute arthritis crisis. Edematous areas of 5-20 cm in diameter, painful and erythematous, appear on the skin.

Pericarditis is rare.

There is emotional lability during attacks.

This condition disappears spontaneously and can recur annually or weekly. In women it is associated with menstruation.

Paraclinical examinations

There is no specific diagnostic test.

During the attack:

Blood. Leukocytosis (15,000 or more). Neutrophilia and moderate anemia. Increased erythrocyte sedimentation rate. Increase in glycoprotein, globulin 2 and plasma fibrinogen. Protein C reactive, haptoglobin, ceruloplasmin. Normal plasma lipids, which allows it to be differentiated from familial hyperlipidemia.

Pleural or peritoneal fluid. Serofibrinous with polynuclear cells up to one million per cubic millimeter.

Radiology. Mild gaseous abdominal distension. Presence of fluid in the abdomen and thorax.

Laparoscopy. Signs of hyperemia in the peritoneum and/or organs; or fibrinous deposits in both sites. Adhesions when there have been several attacks.

Kidney biopsy. Progressive lesion with amyloidosis (40%) or chronic glomerulonephritis.

Syndromogenesis and etiology

Different theories try to explain this syndrome:

1. Syndromic manifestation of palindromic rheumatism.
2. Additional component of chronic rheumatic polyserositis.
3. Decrease in liver glycogen content associated with compensatory hyperadrenalemia.
4. Epileptic equivalent.
5. Present or hidden viral infection.
6. Another hypothesis states that patients are homozygous for a mutant allele of one of the lipocortin genes, which determines deficiency or abnormal production of the lipocortin protein. This deficiency would lead to a loss of eedback inhibition and an increase in the release of arachidonic acid, a precursor of powerful mediators of inflammation; increase in the generation of prostaglandins, leukotrienes and other inflammatory mediators, by granulocytes, which activate phospholipase A2. Lipocortin may be especially critical during stress.

It is transmitted by autosomal recessive inheritance almost exclusively limited to Turks, Armenians, Arabs and Jews. The gene responsible appears to be on chromosome 17.

Literature

Kastner, D.L.; I. Aksentijevich; L. Gruber, et al.: "Familial Mediterranean fever: a 90 marker exclusion map and evidence for linkage to chromosome 17 (Abstract)". Cytogenet. Cell. Genet., 58:21 15, 1991.

Mamou, H. and R. Cattan La maladie périodique (sur 14 cas personnels dort 8 compliqués de néphropathies)". Sem. Hóp. Paris, 28: 1062-1070, 1952.

Mellinkoff, S.M.; R.W. Snodgrass; AD Schwabe, et al,: "Familial Mediterranean fever. " Ann. Int. Med. 56:171-182, 1962.

Meyerhoff, J.: “Familial Mediterranean fever: Report of a large family, review of the literature, and discussion of the frequency of amyloidosis. Medicine., 59: 66, 1980.

Schve, A.D.: "Familial Mediterranean fever in Armenians. Analysis of 100 cases." Medicine. 53:453, 1974.

Shohat, M., J.K. Korember: A.D. Schwabe; J.I. Rotter: "Hypothesis: Familial Mediterrane an fever, a genetic disorder of the lipocortin family? Am. J. Med. Genet.. 34:163-167,1989.

Siegal, S.: "Benign paroxysmal peritonitis." Ann. Int. Med.,23:1--21, 1945.

Siegal, S.: "Familial paroxysmal polyserositis. Analysis of fifty cases." Amer. J. Med., 36:893-918, 1964.

ABDOMINOTHORACIC SYNDROME

Synonymy

S. thoracoabdominal.

With either of the two names it refers to abdominal or thoracic diseases that produce symptoms in the other region, for example:

1. Chest symptoms due to subphrenic abscess, liver abscess.
2. Abdominal symptoms caused by basal pneumonia, pleural empyema, posterior face infarction, etc.
3. Anginous-type reflex pain with or without manifestations on the ECG, caused by cholelithiasis or distal esophagitis (esophageal colic), etc.

Syndromography

Clinic

Pain of variable characteristics and physical signs depending on the cause that originates it.

Paraclinical examinations

They will depend on previous clinical guidance.

Syndromogenesis and etiology

It is a reflex or distant symptom caused by an irritating thorn located at a greater or lesser distance, which is transmitted neurovegetatively or somatically to neighboring metamers.

SEVERE VIGOROSA ACHALASIA SYNDROME

Synonymy

Dyschalasia syndrome.

Syndromography

Clinic

This syndrome occurs in both sexes. It begins between 12 and 79 years, average 51. There is moderate or marked difficulty swallowing, dysphagia, regurgitation, especially when the patient is reclining or lying down, Intense pain in the anterior part of the chest or back, associated with eating while swallowing or not. It lasts for minutes or hours and disappears spontaneously. It is currently considered esophageal colic. There is weight loss, nervousness and mental disorders at times.

Paraclinical examinations

Radiology. Esophagus: cardiospasm. Marked or moderate dilation. Diffuse spasm. Occasionally: diaphragmatic hernia, stenosis or abnormal contractions. Esophageal motility: poor or absent relaxation after swallowing.
Premature contraction of the lower esophageal sphincter. The amplitude of the contractions generates a pressure greater than 50 cm of water. Methacholine test generally positive.

Endoscopy. Achalasia with dilation (30%), hiatal hernia (15%), esophagitis (12%), spasm of the distal esophagus (10%), normal (24%).

Biopsy. Absence of ganglion cells of the Auerbach plexus (not constant).

Syndromogenesis and etiology

This syndrome represents or is a combination of achalasia syndrome and diffuse esophageal spasm syndrome.

The etiology is unknown. The basic defect is not known.

There is familial esophageal achalasia that is transmitted by autosomal recessive inheritance; some reports in the literature indicate that these patients present microcephaly, keratoconjunctivitis, xerostomia, rheumatoid arthritis or other connective tissue diseases. In one of the families described, the siblings showed: achalasia, hyperpigmentation and low levels of steroids in plasma, however, the majority of cases present with achalasia without other associated defects.

Literature

Moersch, H.J.; C.F. Code and A.M.Olsen: "Dyschalasia of the esophagus'". Collect. Papers Mayo Clinic., 49:19, 1957.
Sanderson, D.R.N.; FH. Ellis; J.ESchlegel, et a.: "Syndrome of vigorous achalasia: clinical and physiological observations." Dis. Chest.. 52:508-5 17, 1967.

TYPE I AEROPHAGIA SYNDROME

Synonymy

S. of the air chamber of the stomach.

Syndromography

Clinic

Belching.

Paraclinical examinations

Radiology. Dilation of the air chamber of the stomach.

Fluoroscopy. Air is observed in the esogagus during respiration.
piration

Syndromogenesis and etiology

Frequent and almost always unconscious swallowing of air that accumulates in the stomach and comes out noisily in the form of belching.

Literature

Danhaf, I.E.: "The clinical gas syndromes, a pathophysiological approach." Ann. N.Y. Acad. Sci., 150:127-140, 1968.

TYPE II AEROPHAGIA SYNDROME

Syndromography

Clinic

It is characterized by belching, gastric distension and flatulence.

Paraclinical examinations

Radiology. Dilation of the air chamber of the stomach.

Fluoroscopy. Air is observed in the esophagus during breathing, and the findings are more marked.

Syndromogenesis and etiology

Frequent and almost always unconscious swallowing of air that accumulates in the stomach and comes out noisily in the form of belching. More frequent and longer belching.

bibliography

Danhaf, IE.: “The clinical gas syndromes, a pathophysiological approach." Ann. N.Y. Acad. Sci., 150:127-140, 1968.

TYPE II AEROPHAGIA SYNDROME

Syndromography

Clínic

Massive gastric distention. Respiratory difficulty. Abdomen tympanic to percussion.

Paraclinical examinations

Radiology. Dilation of the air chamber of the stomach.

Fluoroscopy. Air is observed in the esophagus during respiration. piration

Syndromogenesis and etiology

Marked swallowing of air with a dilated and atonic stomach. Frequent in the postoperative period of abdominal surgery.

Literature

Danhaf, I.E.: "The clinical gas syndromes, a pathophysiological approach." Ann. N.Y. Academic Sci., 150:127-140, 1968.

APHTOSIS SYNDROME

Synonymy

Mikulicz thrush. S. de Zahorsky. herpetic angina. Herpangina. cancerous ulcer.

Syndromography

Clinic

This syndrome occurs in both sexes before puberty; It predominates in women after that time. It begins from the first decade to the third, declining thereafter.

It is characterized by small red macules on the oral mucosa that rupture quickly and cause very painful ulcers.
Rare genital location.

Very variable evolution; The lesions return between 7-10 days without leaving traces. They recur periodically.

Paraclinical examinations

Anatomical injuries. Early lymphocytic infiltration around the lobules and ducts of the salivary glands, without previous vascular changes.

Syndromogenesis and etiology

The clinical type called herpangina is due to varieties of the Coxsackie A virus. Other types of aphthosis can be produced by different causes or have an unknown etiology.

Literature

Graykowski, E.A.; M.F. Barile: W.B. Lee, et al.: *Recurrent aphthous stomatits. Clinical therapheutics, histopathologic and hypersensitivity aspects". J.A.M.A., 196:637-644, 1966.
Zahorsky, J.: "Herpangina." Arch. Pediat. N.Y., 41:181-184,1924.

ALBATROS SYNDROME

Syndromography

Clinic

In 1778 Samuel Taylor Coleridge wrote "Rhyme of the Ancient Mariner," a poem that alludes to a young sailor who shot an albatross that was following his ship. Due to this inhuman act against one of God's creatures, all his comrades on board died and he was forced to sail alone through the stormy sea of life with the albatross hanging around his neck until he once again lavished love, reverence and respect on all of God's creatures. In 1967, Johnstone et al used the term albatross syndrome to describe a group of post-gastrectomized patients.

This syndrome appears in post-gastrectomy patients incapacitated by chronic complications after this operation and who have a history of significant personality disorders and a precarious negative relationship with the environmental sectors of work and family. Generally, it occurs in women under 40 years of age in whom the ulcer disease is minimal but the symptoms are maximum. Although the result of the operation may be very satisfactory in the sense of control of the ulcer disease, the patient may be incapacitated by the influence of normal postoperative physiological disorders that he cannot accept or manipulate.

The clinical manifestations of the syndrome are postoperative diarrhea; symptoms such as bloating, fatigue, palpitations and weakness related to rapid gastric emptying. Pain similar to that the patient had before being operated on, nausea and vomiting frequently. Malnutrition and weight loss. Reinterventions do not solve the problem, rather they aggravate it.

The most modern methods for the surgical treatment of ulcers may modify the frequency of postgastrectomy complications, but they will certainly not alter that of albatross syndrome, which are more directly related to the selection of the patient than to that of the surgeon or surgical method.

The psychopathic personality of the majority of patients relates them to Münchausen syndrome and other similar ones that have a complex personality.

Syndromogenesis and etiology

It occurs in stomach surgery patients who have a history of significant personality disorders mentioned above, and abnormal psychic reactions.

Literature

Johnstone, F.R.; I.B. Holubisky; H.T. Debas: “Postgastrectomy problems in patients with personality defects: The albatros syndrome". Canad. Med. Assoc. J.., 96: 1559-1564. 1967.
Krudren, K.B.: "Albatross syndrome: how to prevent it." Clin. Quir. Norteam., 5: 923-926, 1979.

CYCLIC ALLERGIC SYNDROME

Syndromography

Clínica

It is characterized by a condition that recedes and reappears cyclically: digestive symptoms, anorexia, abdominal pain and vomiting sometimes. General symptoms, fever and coryza. allergic asthma.

Syndromogenesis and etiology

This syndrome is due to allergic-type immunological mechanisms and can be triggered by different allergens.

SPLENIC ANGLE SYNDROME

Synonymy

S. de Payr. (See irritable bowel syndrome.)

Syndromography

Clinic

It is present in about 20% of patients with irritable bowel syndrome. Generally postprandial. Pain in the left hypochondrium that can radiate to the precordial region, left thoracic region or back, neck and left arm. Sometimes tachycardia and dyspnea. Sometimes pressure or pain in the rectum.

Physical examination. Abdominal distension (occasional) or palpation of a spastic colon.

Paraclinical examinations

Radiology. Gas accumulation and localized distention in the splenic flexure of the colon on plain radiography.

The enema colon shows the splenic flexure between the diaphragm, stomach and spleen, and often shows a very pronounced kink.

Literature

Bockus, H.L.: Gastroenterology. ed. 3, Philadelphia, Saunders, 1994.

Payr, E.: "Ueber eine eigentümliche, durch abnorm starke Klickngе und Adhasionen bedinge gucartige Stenose del Flexura lienalis und heapatice coli." Verh. Deut. Keng. Inn. Med., 27:276-305, 1910.

INTESTINAL MOTILITY ABNORMALITY SYNDROME

Syndromography

Clínica

It is observed in old age. It is characterized by distention of the colon. Painful sensation in the splenic and hepatic angles of the colon. Decreased flatulence.

Paraclinical examinations

Radiology. Motility from the stomach to the cecum is normal, but in the cecum it is decreased.

Syndromogenesis and etiology

It is due to colonic hypomotility.

Literature

Danhaf, I.E.: "The clinical gas syndromes, a pathophysiological approach. Ann. N.Y. Acad. Sci., 150:127-140, 1968.

INTESTINAL MOTILITY ABNORMALITY SYNDROME II

Syndromography

Clinic

It is characterized by borborygmus and sensation of bubbles, occasionally diarrhea. Abdominal distention and flatulence.

Paraclinical examinations

Radiology. Gas is observed in the small intestine with a decrease in transit time, as can be verified with carmine and/or barium.

Motility test. Increased colonic motility.

Syndromogenesis and etiology

It is due to intestinal hypermotility. Increased secretion of mucus.

Bibliography

Danhaf, L.E.: “The clinical gas syndromes, a pathophysiological approach." Ann. N.Y. Academic Sci., 150:127-140, 1968.

ABDOMINAL STROKE SYNDROME

Synonymy

Spontaneous massive intraperitoneal hemorrhage. S. of intra-abdominal apoplexy. S. of mesenteric or subperitoneal hemorrhage.

Syndromography

Clinic

Preferably in men with arteriosclerotic hypertension. It has an acute onset and is characterized by progressive abdominal pain accompanied by nausea. Vomiting does not relieve the pain.

If hemorrhage persists, it leads to collapse and shock.

Physical examination. Signs of peritoneal irritation: distension, pain, muscle contracture.

Syndromogenesis and etiology

Anatomical injury: spontaneous rupture of an intra-abdominal vessel without history of trauma or aneurysm, atherosclerotic lesions in young adults.

The rupture of a small, localized aneurysm has also been suggested.

Literature

Bockus, HL.: Gastroenterology. ed.3, vol. 3, Philadelphia, Saunders, 1980.

Cushman, G.E. and A.R. Kilgore: "The syndrome of mesenteric or subperitoneal hemorrhage (abdominal apoplexy)." Ann. Surg.. 114:672-681, 1941.

SUPERIOR MESENTERIC ARTERY SYNDROME

Synonymy

S. arteriomesenteric-duodenal obstruction. S. of gastromesenteric ileus. Aortomesenteric impingement. S. de Wilkie. S. of vascular compression of the duodenum.

Syndromography

Clinic

Acute form. Sensation of postprandial gastric distension. Epigastric cramps accompanied by vomiting, with bile content, especially in the erect position. Sensation of heaviness or oppression in the epigastrium caused by dilation of the stomach. Fear of eating and weight loss.

Subacute-chronic form. Periodic abdominal pain especially after meals, cyclical postprandial vomiting.

Paraclinical examinations

Stomach and duodenum radiology. Hyperdistention of the stomach and duodenum; absence of air in the distal duodenum.

If barium is administered: dilated stomach, open pylorus, dilation of the initial portion of the duodenum. This dilation is evident and can measure 4.8 cm, the normal is 0.95 cm.

If the patient is placed in a prone position on the right flank or supported on the elbows and knees, the contents are emptied and the symptoms are relieved.

Syndromogenesis and etiology

It is due to intermittent compression of the third portion of the duodenum by the superior mesenteric artery at its origin. The two most important factors in the production of this syndrome are: the narrower angle of the mesenteric artery and the shortness of its length from the exit of the artery to crossing the third portion of the duodenum. It may be associated with increased tension in the root of the mesentery of congenital origin.

Rapid weight loss may contribute.

Literature

Choi, S.H. and F.A. Pfalzer Jr.: "Superior mesenteric artery syndrome'" N.Y. St. J. Med., 76:986-988, 1976.

Wilkie, D.P.D.: "The blood supply of the duodenum. With special reference to the supraduodenal artery." South. Gynec. Obst., 13:399-405, 1911.

AFERENT LOOP SYNDROME

Synonymy

S. of obstruction of the gastrojejunal loop. S. of the iatrogenesis of the afferent loop. Stagnant Loop Syndromes.

Syndromography

Clinic

Anorexia, nausea, postprandial fullness, pain in the upper abdomen followed by bilious vomiting without food remains.

Fatty foods precipitate vomiting. profuse diarrhea, fever. These symptoms can be light, moderate or intense with weight loss and intolerance to moderate foods.

Physical examination. Abdominal distention, palpable mass, progressive weight loss.

Paraclinical examinations

Radiology. Dilation of the proximal jejunal segment. Peristaltic waves fight against an obstacle. Dilution of barium by bile and pancreatic juices. Jejunogastric regurgitation.

Retention of contrast medium in the proximal jejunum.

Blood. Megaloblastic anemia.

Anatomical injuries. The afferent loop may present partial volvulus, herniation, adhesions, or be too short or too long. Signs of inflammation that can lead to necrosis and perforation with peritonitis.

Syndromogenesis and etiology

A stasis of secretions and foods occurs that favors bacterial overgrowth with splitting of bile salts, which are largely the cause of the symptoms. It behaves like other overgrowth syndromes in the intestine or blind loop syndrome.

It is due to surgical iatrogenesis during partial or subtotal Billroth II gastrectomy. Less frequently it is due to postoperative adhesions in the suture area.

Literature

Schjonsby, H.; J.R. Halvorsen, et al,: "Stagnant loop syndrome in patients with continent ileostomy (intra-abdominal ileal reservoir)". Gut, 18:795-799, 1977.

Tabaqchali, S.; J. Hatzioannou and C.C. Booth: Bile salt deconjugation and steatorrhea in patients with the stagnant loop syndrome." Lancet, 2:12-16, 1968.

BLIND LOOP SYNDROME

Synonymy

S. from the blind bag. S. of the contaminated handle. S. of abnormal bacterial growth. S. of the stagnant handle. S. of intestinal stasis. S. of the complicated side-lateral anastomosis.

Syndromography

Clinic

This syndrome generally begins years after surgery with side-to-side anastomosis. It is characterized by weakness. Inability to gain weight. Intermittent diarrhea. Painful abdominal cramp. Abdominal distention, nausea and vomiting. Multiple vitamin deficiency symptoms: A, K, D, Malabsorption with steatorrhea, this causes electrolyte deficiency, glossitis, paresthesias and sometimes tetanic symptoms, osteoporosis.

These symptoms may occur alone or in combination.

Physical examination. Paleness, abdominal discomfort and/or palpable soft handle.

Paraclinical examinations

Blood. Megaloblastic anemia.

Radiology. Demonstration with contrast media of the loops in the intestine.

Anatomical injuries. Dilation of the afferent and efferent loops. Hypertrophy, edema and ulceration of the mucosa.

Syndromogenesis and etiology

The fundamental thing is dysbacteriosis and the bacterial overgrowth that causes:

1. Excessive deconjugation of bile acids with subsequent micellar decrease.
2. Consumption, degradation or conjugation of vitamin B12 and other vitamins, carbohydrates and proteins.

Other causes in non-operated cases are: diverticula, intestinal stenosis, scleroderma, adhesions with subocclusion, afferent loop syndrome, etc.

Bibliography

Botsfor, T.W. and A.B. Gazzaniga: “Blind pouch syndrome, a complication of side to side intestinal anastomosis." Amer. J. Surg., 113:486-490. 1967.

EFFERENT LOOP SYNDROME (GASTROJEJUNOSTOMY)

Syndromography

Clinic

This syndrome begins 8-10 days after the patient has surgery. It is characterized by bile-tinged vomiting with a feeling of discomfort and nausea in the epigastrium.

Paraclinical examinations

Radiology. Poor filling of the efferent segment that is flaccid 2-10 cm below the gastroenterostomy site.
Marked slowness in emptying. Normal appearing mucosa.
Absence of generalized paralytic ileus.

Syndromogenesis and etiology

It is a functional disorder of the efferent jejunal loop that causes a delay in emptying. This situation can last a few days or several weeks.

Literature

Bodon, G.R., et al.: "The gastrojejunostomy efferent loop syndrome." Surg. Obstet. Gynecol., 134:777, 1972.
Golden, R.: "Functional obstruction of the efferent loop of jejunum following partial gastrectomy." J.A.M.A., 148:721, 1952.
Lefebure, F.: "La pathogenie de l un anse efferente current" Hepatho- - Gastroenterol. d'Hotel Dieu, l:256, 1965.

BANTI SYNDROME

Synonymy

S. of congestive splenomegaly. Fibrocongestive splenomegaly. Hepatosplenome gallium. Cirrhotic splenomegaly.
S. of spleen-liver. Splenic anemia. Banti disease.
Hepatolienal fibrosis.

Syndromography

Clínic

This syndrome is more common in patients under 35 years of age. Its onset is insidious or due to hematemesis.

It is characterized by weakness, flatulence, diarrhea, signs of slow digestion and abdominal discomfort. Epistaxis (30%).

It was described by Banti and other clinicians considering three stages:

I. Anemic stage. Painful crises in the splenic region, sensation of oppression in the epigastrium, digestive disorders. Splenomegaly of hard consistency that increases progressively. Hypochromic anemia with moderate leukopenia, thrombopenia and relative or absolute lymphopenia. Recurrent digestive bleeding.

II. Transition stage. Sclerocutaneous subicterus. Urobilinuria.

II. Ascitic stage. Liver shrinkage, ascites, pigmentation tanning of the skin, increased jaundice, collateral circulation, progressive cachexia and death.

Paraclinical examinations

Blood. Normocytic type anemia, leukopenia and thrombocytopenia with hyperplasia of the three systems in the bone marrow (hypersplenism). In many cases alterations in liver function tests, especially bromosulfthalein.

Radiology. Esophageal varices. Splenoportography shows obstruction of the portal system, of intrahepatic cause due to cirrhosis or extrahepatic.

Syndromogenesis and etiology

Intrahepatic portal hypertension (caused by cirrhosis or schistosome) or extrahepatic (portal thrombosis, compression, aneurysm).

Literature

Banti, G.: “Dell'Anemia splenica”. Arch. Scuola Anat. Patol., Firenze, 2:53-122, 1883.

BARCLAY-BARON SYNDROME

Syndromography

This syndrome is formed by indirect signs of duodenal ulcer:

1. Hypersecretion of gastric juice.
2. Gastric hypertonia characterized by rapid emptying of barium at the beginning and slow after the first moments.

Syndromogenesis and etiology

Gastric hypersecretion is a reflex phenomenon produced by duodenal ulcer.

Hypertonia is due to a gastric motility disorder for the same reason.

Gastrointestinal hormones are probably involved in both, especially gastrin.

BARD-PICK SYNDROME

Synonymy

S. of pancreatic malignancy. S. de Courvoisier-Terrier.

Syndromography

Clinic

I start preferably between the fourth and fifth decades.
Male-female ratio 2:1. Symptoms depend on location

of the shift. The typical triad: progressive, afebrile, painless jaundice. Vesiculomegaly (Courvoisier sign). Progressive and rapid cachexia. Also: anorexia, weight loss, steatorrhea. This triad is present when it affects the head of the pancreas especially and corresponds to what was described by Bard and Pick.

In other locations of the neoplasm, severe pain is characteristic, but of undefined characteristics, which frequently affects the back, nausea, flatulence and sometimes vomiting. Absence of jaundice during most of its evolution.

Physical examination. Jaundice, weight loss. Sometimes supraclavicular sentinel lymph node. Palpable vesicle. Ascites especially when located in the body and

It is very important to keep in mind that Bard-Pick syndrome is frequently accompanied by psychological manifestations that confuse the doctor, especially in pancreatic body neoplasia.

Paraclinical examinations

Blood. Anemia. Increased lipase and amylase. Conjugated hyperbilirubinemia.

Radiology. Stomach compression. Widening and compression of the duodenal frame in advanced cases.

Ultrasound and computed tomnography. Shows the tumor lesion.

Laparoscopy. It confirms an increase in the gallbladder, green hepatomegaly, and the tumor lesion can be observed and biopsied in most cases.

Splenomegaly when the splenic vein is compressed.

Duodenoscopy and retrograde cholangiography. Cannulation of the sphincter of Oddi when possible shows: dilation of the bile and pancreatic ducts.

Laparoscopic cholecystocholangiography or percutaneous cholangiography. It is much more useful in cases where the previous examination fails or the lesion is advanced.

Death almost always occurs within a year of the onset of symptoms.

Syndromogenesis and etiology

This syndrome is due to a malignant neoplasm of the pancreas, usually a carcinoma.

Bibliography

Bard, L. and A. Pick: “Contribution a l'etude clinique et anatomo athologique du Cancer primitif du pancreas." Rev. Méd., Paris, 8:257-282, 363-405, 1888.

Berck, E.J. and W.S. Hanbrick: Tumor of pancreas. In H.L. Bockus: Gastroenterology. vol. 3, Philadelphia, Saunders, 1980, pp. 1168-1186.

BARRETT SYNDROME

Synonymy

Barrett's esophagus. Barrett's ulcer. S. of chronic peptic ulcer and esophagitis. Allison-Johnstone anomaly.

Syndromography

Clinic

This syndrome generally occurs in middle age or old age. Recurrent retrosternal pain with heartburn. The pain may radiate to the neck, scapular region or arms.
It almost always occurs after eating hot or cold foods and in the supine position. Esophageal regurgitation.

In advanced cases there is dysphagia, hematemesis and/or melena.

Paraclinical examinations

Radiology. Small niche in the wall of the esophagus with absent or atypical folds above the niche. Stenosis due to edema.

Esophagoscopy. Crater, poor fold development, edema and signs of inflammation. Leukoplakia and sometimes signs of hemorrhage. These lesions are frequently located in the middle or lower esophagus.

Biopsy. Absence of malignancy, only chronic inflammatory elements. The most specific is the replacement of the squamous epithelium of the esophagus with columnar epithelium. It can evolve towards malignancy (2-5%) and must be monitored periodically.

Syndromogenesis and etiology

The fundamental mechanism seems to be gastroesophageal reflux and/or chronic lesions that produce a replacement of the stratified squamous epithelium with columnar or cylindrical epithelium that is much less resistant. Hiatal hernia exists in 85% of cases.

It is always associated with ulcerations and/or stenosis of the esophagus, probably caused by the described mechanism caused by a chronic peptic ulcer of the stomach.

Literature

Barrett, N.R.: "Chronic peptic ulcer of the esophagus and esophagitis." Br. J. Surg., 38:175-182, 1950.

Rentsch, J.; H. Holz; R. Baszus and FK. Kosling: "Barrett-Syndrome." Med. Klin., 74:437-441, 1979.

BÁRSONY-POLGÁR SYNDROME

Synonymy

S. de Bársony-Teschendorf. Segmental spasm of the esophagus. Multiple esophageal spasm.

Syndromography

Clinic

Intermittent, paroxysmal dysphagia, with regurgitation of saliva and mucus accompanied by intense retrosternal pain due to crises.

Sometimes the patient cannot swallow for an hour.

It is associated with chronic gastric and duodenal ulcers, recurrent colitis, biliary dyskinesia, cholecystitis and pericholecystitis, cholelithiasis, and coronary spasm.

Paraclinical examinations

Esophageal motility. Abnormal contractions and increased intraesophageal pressure; repeated, synchronous contractions with an increase in their amplitude and duration.

Radiology. Tertiary contractions: multiple areas with saccular dilations and narrowings. The contractions can be in a single segment or also waves with marked peristalsis, a "string of pearls" esophagus.

Syndromogenesis and etiology

It is considered a functional motor disorder. It may be a neurovegetative disorder, a tetanoid condition, or a visceral reflex on the vagus nerve.

The etiology is unknown.

Literature

Bársony, T. and A. Polgár: “Synptomolose und funktionelle Speiseröhrendivertikel.” Fortschr. Röntgen., 36:593, 1927.
Bennet, J.R., et al.: "Diffuse esophageal spam: A disorder with more than one cause." Gastroenterology, 59:273, 1970.
Teschendorf, W.: "Die Röntgenuntersunchung der Speiseröhre." Ergeb. Med. Strahl., 3:175, 1928.

BOERHAAVE SYNDROME

Synonymy

S. of spontaneous laceration of the esophagus.

Syndromography

Clinic

Prevalence in men (5:1), more common in the fifth to sixth decades. It occurs when great efforts are made to vomit, in the form of transfixing pain in the thorax and epigastrium, progressive dyspnea with tachypnea, expulsion of small amounts of blood through the mouth, more rarely intense hematemesis. Feeling of anguish and shock.

Physical examination. Signs of hypotension, tachycardia and especially subcutaneous emphysema, especially in the cervical region and face.

Paraclinical examinations

Simple radiology. Demonstrates pneumomediastinum.

Hemogram. Decrease in hematocrit and anemia, if the bleeding is significant.

Electrocardiogram. Absence of signs of ischemia myocardial.

Esophagoscopy. It allows you to visualize the perforation, as well as its extent and whether the bleeding is significant. A minimum of air should be used to avoid increasing pneumomediastinum and/or pneumothorax.

Anatomical injuries. Longitudinal linear perforation almost always on the dorsal aspect of the lower esophagus. Hydrothorax and pneumothorax.

Syndromogenesis and etiology

It is a laceration or tear of the wall of the esophagus that includes all layers. The mechanism is due to the increase in intra-abdominal and intraesophageal pressure almost always during strong retching or vomiting.

Literature

Boerhaave, H.: Atrocis, nec descripti prius, morbi historia. Secundum medicae artis leges conscripta. Lugduni Batavorum, Boutesteniana, 1724.
Bruno, M.S.; W.R. Grier and W.B. Ober: "Spontaneous laceration and rupture of esophagus and stomach, Mallory-Weiss syndrome, Boerhaave syndrome, and their variants." Arch. Int. Med., 12:574-583, 1963.
Patton. ACE. and D.W. Lawson: Reevaluation of the Boerhaave syndrome. A review of fourteen cases". Am. J. Surg. 137: 560, 1979

BÖÖK SYNDROME

Synonymy

S. of premolar aplasia-hyperhidrosis-canicie (PHC).

Syndromography

Clinic

This syndrome is characterized by premolar aplasia (P), manifest functional hyperhidrosis (H) and premature gray hair (C) that appears between 6 and 23 years of age. Hyperhidrosis is observed on the palms of the hands and soles of the feet, accompanied by palmoplantar keratosis. There is hypodontia with premolar aplasia.

Syndromogenesis and etiology

It is transmitted by highly penetrant autosomal dominant inheritance. In 1950, Böök reported 25 affected people in four generations of a Swedish family.

Literature

Böök, J.A.: "Clinical and genetic studies of hypodontia. I. Premolar aplasia, hyperhidrosis and premature canities. A new hereditary syndrome in man. " Am. J. Med. Genet., 2:240-263, 1950.

BOUVERET SYNDROME

Syndromography

Clinic

Colicky abdominal pain generally in the epigastrium and/or right hypochondrium, accompanied by bilioalimentary nausea and vomiting, which relieves the pain. Vomiting is frequent and prevents feeding. Sometimes there are signs of peritoneal irritation on physical examination.

Paraclinical examinations

Ultrasound. It may show the stone in the pylorus or in the first or second portion of the duodenum.

Radiology. It allows the stone to be seen, large, usually single if it is opaque to X-rays, otherwise the administration of barium in a thin layer can demonstrate it. Air is also observed in the bile ducts, an important sign to recognize the cause of the occlusive condition.

Panendoscopy. It directly visualizes the stone and allows its extraction or destruction, thus avoiding surgery in most cases.

Syndromogenesis and etiology

The first two patients diagnosed preoperatively with this syndrome were due to Bouveret (1896), although in autopsy it had been described by Bonnet in 1841.

It is produced by the migration and locking of a large stone from the gallbladder to the stomach or duodenum through a spontaneous fistula, either at the starting point of the gallbladder or a gastroduodenal ulcer.

When the stone is not so large, occlusion occurs in the terminal ileum, causing gallstone ileus syndrome.

Literature

Bedogni, G.; S. Contini; M. Meinero, et al,: “Pyloroduodenal obstruction due to a biliary stone (Bouveret's syndrome) Managed by endoscopic extraction." Gastroint. Endosc., 31: 36, 1986.
Ryska, M.; M. Prokhazka; J. Skala, et al: "Gallstone Pyloro duodenal obstruction. Fiberscopic diagnosis." Chir Act. Scand., 151:191, 1985.

Simonian, S.J: "Gallstone obstruction of the duodenal bulb." Lancet, 1:893-894, 1968.

BRENNEMANN SYNDROME

Synonymy

Acute mesenteric lymphadenitis. Mesenteric adenitis.

Syndromography

Clinic

It is characterized by abdominal pain, generally in the right lower quadrant, nausea, vomiting and fever.

It occurs more frequently in children under 15 years of age during an upper respiratory tract infection.

Physical examination. Pain on palpation of the right lower quadrant with signs of peritoneal irritation evidenced by the Blumberg maneuver. Easy to confuse with acute appendicitis.

Nasopharyngeal infection, especially of the tonsils.

Paraclinical examinations

Blood. The leukogram is not conclusive, in many cases it reveals an increase in lymphomonocytes (virocytes).

Laparoscopy. It must be performed urgently and is the most accurate method for diagnosis. It allows us to check the normality of the appendix and in many cases see the increase in the mesenteric lymph nodes.

Anatomical injuries. Adenitis of the mesenteric and retroperitoneal

Syndromogenesis and etiology

Brennemann syndrome is due to a viral infection (adenovirus and others) that causes a marked increase in the size of the abdominal lymph nodes, which causes pain and clinical symptoms.

Literature

Brennemann, J.: The abdominal pain of throat infections." Ame. J. Dis. Child.. 22:493-499, 1921.

BUDD-CHIARI SYNDROME

Synonymy

S. of Chiari. Chiari disease. Thrombosis of the hepatic veins. S. de Rokitansky-Chiari. Budd's disease.

Syndromography

Clinic

More common in men. Acute form: abrupt epigastric pain with nausea, vomiting, hematemesis, coma and shock.

Chronic form: symptoms and signs develop gradually and/or intermittently, pain in the right upper quadrant, liver enlargement, rarely splenomegaly.

Physical examination. Progressively increasing hepatomegaly.

Ascites and edema of the lower limbs. Collateral circulation. Discrete splenomegaly. Occasionally jaundice and/or gastrointestinal bleeding due to esophageal varices.

Paraclinical examinations

Liver tests. Moderate increase in bilirubin. Elevated alkaline phosphatase. Elevated transaminase.

Determination of portal pressure, isotopic scintigraphy of the liver, splenoportography. They allow the diagnosis to be established.

Radiology. Shows esophageal varices.

Selective arteriography of the hepatic artery and/or splenic artery. They help in the diagnosis.

Cavography. It shows stasis and sometimes thrombus in addition to collateral circulation if splenoportography is performed.

Laparoscopy with liver biopsy. They are the most accurate methods, not only for diagnosis but for assessing the stage of the liver.

Syndromogenesis and etiology

It is due to acute or chronic obstruction of the suprahepatic veins by thrombus or tumor. This syndrome is observed more frequently after the use of oral contraceptives. Congenital venous malformation, traumatic or mechanical factors and hematological alterations have been suggested: sickle cell disease, leukemia, polycythemia. Endophlebitis of the suprahepatic veins, bile duct infections.

Whatever the cause of the occlusion, venous stasis is created in the liver leading to hepatomegaly with fatty infiltration, edema and centrilobular fibrosis. Hepatic pseudocirrhosis similar to chronic cardiac liver or that seen in the peri-constrictive carditis.

The etiology is unknown in two out of three patients.

Literature

Budd, G.: On Diseases of the Liver. London, J. Churchill, 1852.

Chiari, H.: "Über die selbständige Phlebitis obliterans der Haupstämme der Venae hepaticae als Todesursache." Beitr. Path. Anat., 26:1-17, 1899.

BURNETT SYNDROME

Synonymy

S. milk and alkaline. S. of milk drinkers. S. of pathological calcifications. S. of alkalosis. S. of dietary hypercalcemia.

Syndromography

Clinic

There are three forms of clinical presentation in milk-alkali syndrome: acute, subacute and chronic. The latter is what is known as Burnett syndrome, which presents kidney complications due to calcifications. The acute form presents with hypercalcemia and alkalosis a few days after ingesting milk and alkali, and is characterized by weakness, myalgia, irritability and apathy, with kidney function disorders.

This condition disappears when treatment is stopped.

Other symptoms are: weakness, headache, dizziness, nausea and vomiting, depression and anorexia.

Physical examination. Calcinosis manifested by calcareous deposits in the conjunctival angle and sometimes in the cornea, bands of keratitis in the eyes, ataxia and mental confusion.

Paraclinical examinations

Blood. Hypercalcemia. Hyperazotemia. Mild alkalosis.

Urine. Normal calcium and phosphates.

Radiology. Calcium deposits in different tissues of the body: nephrocalcinosis, coneal calcification, soft tissue calcification.

Syndromogenesis and etiology

Prolonged and/or excessive ingestion of milk and antacids, especially bicarbonate due to gastric lesions: ulcers, gastritis, etc. With the use of non-absorbable antacids, the frequency has decreased.

It is the result of chronic ingestion of calcium and alkali in susceptible patients. The perpetuity of the cycle: mild hypercalcemia - bicarbonate retention - alkalosis - renal calcium retention - severe hypercalcemia, makes the process irreversible.

There is a certain predisposition to present this syndrome, probably due to disorders in the absorption of calcium and/or parathyroidism.

Literature

Burnett, C.H.; R.R. Commons; F. Albright and J.E. Howard: "Hypercalcemia without hypercalciuria or hypophosphatemia, calcinosis and renal insufficiency: A syndrome following prolonged intake of milk and alkali." N. Engl. J. Med., 240:787-794, 1949.

Orwoll, E.S.: "The milk-alkali syndrome: Current concepts." Ann. Intern. Med. 97:242, 1982.

CANAL PUDENDU SYNDROME

Syndromography

Clínic

It occurs in female patients and is characterized by anal pain, which does not respond to analgesics nor is it triggered or relieved by defecation, accompanied by perianal numbness and itching. In some patients there is incontinence of liquid stools and gas.

Some point out that the symptoms appeared after difficult births and others complain of severe straining to evacuate their bowels.

Physical examination. A rectal examination shows that the anal reflex is very weak or imperceptible, and the pressure of the rectal neck when voluntarily squeezing is low.

Paraclinical examinations

Electromyogram. This test, performed on the external anal sphincter and levator ani, is reduced to compression and/or stimulation.

Syndromogenesis and etiology

This syndrome has been recently described and is due to compression of the pudendal nerve in the pudendal canal. Surgery performed at this level frees the nerve from its compression and the patient from her symptoms.

Literature

Shafik, A.: "Pudendal canal syndrome. New syndrome." Colo- - Proctology, VIII (2), 1992.

HEREDITARY NON-POLYPOSIC COLORECTAL CANCER SYNDROME

Synonymy

S. HNPCC (hereditary non-polyposic colorectal cancer).
S. de Lynch I and II. S. of familial cancer.

Syndromography

Clinic

Colorectal cancer (CRC) occurs at an early age, around 44 years of age, usually in the right colon (70%).

An excess of synchronous and metachronous colorectal tumors is common (45% ten years after hemicolectomy or resection of the affected segment).

They do not develop from polyps, but from adenomas similar to those of the general population.

In Linch I type, malignant tumors are limited to the colon or specific sites in this organ.

In Lynch type II, in addition to colorectal cancer, there is a high frequency of endometrial cancer, transitional cell cancer of the urethra and renal pelvis, and adenocarcinomas of the stomach, small intestine, ovary, pancreas, and biliary tract. But it does not always occur in specific places like the other type.

Syndromogenesis and etiology

It is seen in Caucasians, blacks and Asians and only once in Native American Indians (in a Navajo family).

It is a disease with autosomal dominant inheritance.

Carriers have a high risk of acquiring colorectal cancer with an estimated penetrance of 85%.

Differential diagnosis.

When considering the diagnosis of HNPCC it is important to ex-include:

1. Familial polyposis adenomatosis (familial adenomatous polyposis FAP).
2. Hereditary flat adenoma syndrome (HFAS).
3. Hamartomatous polyposis syndromes (Peutz-Jeghers syndrome, familial juvenile polyposis syndrome).
4. Family aggregates with colorectal cancer without sufficient criteria to be considered hereditary.
5. Non-multiple hereditary polyposis carcinoma.
6. Familial chronic ulcerative colitis.

The difference between FAP and HNPCC is easy, due to the abundance of adenomas in the former. However, the attenuated variant of FAP (HFAS) may have very few adenomas. In these cases, the presence of upper gastrointestinal lesions can help, since polyps of the fundic glands are characteristic of HFAS and are not found in HNPCC. Periampullary adenomas and carcinomas that are not typical of HNPCC may also occur in HFAS.

Hamartomatous polyposis have specific pathological characteristics that allow them to be differentiated from HNPCC.

The genetic basis of HNPCC has been proven by linkage studies involving chromosome 2p in some families and 3p in others. Mutations on either chromosome are present in 90% of known HNPCC families.

HNPCC is considered to be due to a constitutional mutation in the base mismatch repair genes during DNA replication.

Literature

Lynch, H.T. and J. Lynch: "Genetics natural history, surveillance management and gene mapping in the Lynch syndrome." Pathol. Biol., 43(3):151-158, 1995.

Thomas, G. and S. Olschwang: "Genetic predispositions to colorectal cancer." Pathol. Biol., 43(3):159-164, 1995.

CARCINOID SYNDROME

Synonymy

S. de Thorson-Biörck. S. of malignant carcinoid. S. of argentafinoma. S. of intestinal carcinoid. Carcinoidosis. Cassidy S. S. de Hedinger. S. de Scholte.

Syndromography

Clínic

Vasomotor phenomena of redness (hot flush) and/or cyanosis, episodic, located especially in the head and neck. This symptom can be triggered by effort, emotion, ingestion of food and/or alcohol. If it is prolonged, it is accompanied by lacrimation and periorbital edema.

Asthmatic crisis. Explosive diarrhea that accompanies hot flashes with abdominal cramps and borborygmi. In other cases persistent chronic diarrhea. Bulimic attacks with spontaneous hypoglycemia.

Physical examination. In some patients, violent telangiectasia, especially on the face (malar region) and neck.

Cardiovascular symptoms that can lead to right-sided insufficiency, caused by: endocardial fibrosis, pulmonary stenosis, tricuspid insufficiency. Murmur and thrill in the right ventricular region. Jugular engorgement, hepatomegaly, edema and ascites occasionally. Pelagroid dermatitis and alterations that simulate scleroderma, in the lower limbs.

Paraclinical examinations

Urine. There is an overproduction of 5-hydroxyin-doles with an increase in 5-hydroxy-3-indoleacetic acid in 24-h urine (should not exceed 9 mg). In positive cases it can reach up to 1,000 mg. However, it remains within normal limits in other types of hot flashes, which allows a differential diagnosis to be made with these, which are due to a systemic activation of mast cells including mastocytosis and those that accompany the postmenopausal state.

Radiology. The small intestine is the site where the tumor is most frequently located and the radiological findings are: in the beginning stages, atypical intramural defect; in advanced stages, lobe or intraluminal tumor. Pulmonary or cardiovascular signs when these structures are affected.

Syndromogenesis and etiology

The symptoms are fundamentally due to the excessive production of serotonin (5-hydroxytryptamine) and other substances by the chromaffin cells of the functioning tumor (Kultschitzky cells). The tumor can arise in the epithelium of the gastrointestinal tract, from the mouth to the anus; even in the epithelium of the bile and pancreatic ducts. More rarely, in the bronchial epithelium it causes metastases to the liver, lymph nodes, lung and bones.

Specifically, it is considered that serotonin is not the cause of hot flashes, since these can also occur due to excess histamine (gastric carcinoid), pentagastrin, catecholamines, isoproterenol, etc.

In relation to cardiac alterations, Melmon and collaborators have isolated from the carcinoid tissue another biologically active substance that can cause all the symptoms of the syndrome, it is an enzyme analogous to kallikrein that releases from the plasma protein (in patients with carcinoid) an active peptide with vasomotor action.

Literature

Biörck, G.; 0. Axén and A. Thorson: "Unusual cyanosis in a boy with congenital pulmonary stenosis and tricuspid insufficiency, fatal outcome after angiocardiography". Amer. Heart J., 44:143- -144, 1952.

Cassidy, M.A.: “Abdominal carcinomatosis with probable adrenal involvement.” Proc. Roy. Soc. Med. London, 24:139, 1930- -1931.

Gardner. B. M. Dollinger: W. Silen: et al.: *Studies of the carcinoid syndrome: its relationship to serotonin, bradykinin, and histamine". Surgery, 61:846-852, 1967.

CAROLI SYNDROME

Synonymy

S. of segmental dilation of the intrahepatic biliary tree.

Syndromography

Clínica

Clinical manifestations can present in very varied forms and have been described in the first months of life up to 60 years of age. The symptoms are: pain in the right hypochondrium, subicterus or jaundice, enlarged liver and fever. Tendency to intrahepatic cholelithiasis, cholangiitis and liver abscesses.

It may be associated with other anomalies: renal cysts, fibroangiomatosis of the bile duct.

Paraclinical examinations

Cholangiography. Saccyform segmental dilations of the intrahepatic biliary tree, the size of which can reach that of a large lemon, and are communicated with the biliary system.

Laparoscopy. Small alterations of the liver surface (inconstant) with irregularities, but without signs of liver cirrhosis.

Syndromogenesis and etiology

Caroli syndrome is due to a congenital anomaly. It differs from polycystic liver because in this case there is no communication of the cysts with the bile duct.

The etiology is unknown.

Literature

Caroli, J. et V. Corcos: Maladies de voies bile intrahepatiques segmentaires. Masson, Paris, 1964.

Hermansen, M. C..RJ. Starshak and St. L. Werlin: "Caroli disease: The diagnostic approach". J. Pediat. 94:879-882, 1979.

CELIA CO SYNDROME

Synonymy

Gluten-induced enteropathy. Infantile sprue. S. de Gee-Herter-Hubner. Non-tropical sprue s.

Syndromography

Clinic

It begins between 8-24 months and is characterized by a basic triad: a, diarrhea with pale, voluminous, fatty and foul-smelling stools; b, bulging abdomen and emaciated buttocks: c, marked anorexia.

Poor development with malnutrition (sometimes leading to edema), stunted height, bad mood, irascibility.

Physical examination. Paleness (anemia), tetany, rickets, hemorrhages.

Paraclinical examinations

Stool. Steatorrhea greater than 3.5 g/24 h.

D-xylose test. Excretion less than 15%.

Intestinal biopsy. Decrease in the number and flattening of villi. Deep crypts.

Radiology. Contrast flocculation (snowfall). Hypotonic loops with wide folds in the intestine.

Syndromogenesis and etiology

The mechanisms that produce the symptoms are due to damage to intestinal cells by gluten. Immune complex deposits have been demonstrated in parietal cells. The syndrome is associated with HLA-DR3 and HLA-DQW2 in all populations studied.

It is transmitted by autosomal dominant inheritance with reduced penetrance or by polygenic inheritance. There is 70% concordance in monozygotic twins.

Literature

Gee, S.: "On the celiac affection." St, Bartholomew's Hosp. Rep. 24, 1888.

Young, W.E. and E.M. Pringle: "110 children with celiac disease 1950-1969." Arch. Dis. Child., 46:421, 1971.

Trier, J.S.: "Celiac sprue." N. Eng. J. Med., 325:1709-1719, 1991.

HYPERMOBILE BLIND SYNDROME

Synonymy

S. of habitual torsion of Klose's cecum. S. of painful mobile cecum.

Syndromography

Clinic

Recurrent crampy abdominal pain located periumbilically or in the right lower quadrant, which is confused with the pain of intestinal obstruction or acute appendicitis.

Physical examination. Pain is noted on palpation.

Paraclinical examinations

Radiology. It shows excessive mobility of the cecum and ascending colon that reaches the umbilical region, with dilation and stasis.

Laparoscopy. Highly vascularized peritoneal insertion in the external part of the ascending colon. Enlargement of lymph nodes. Sometimes retrocecal appendix.

Syndromogenesis and etiology

It is an anomaly that is characterized by an increase in the length of the omentum that fixes the cecum and the lower part of the ascending colon, which allows great mobility, kinking and pulling of the meso that can reach torsion with the consequent occlusive and vascular condition.

Literature

Bruns. HA. et al.: «The mobile cecum". Progr. Pediatr. Radiol. 2:352, 1969.

Nicole, R.:Über das Coecum-mobile sydrom bei Kinderm". Ann. Paediatr. (Basel), 183:346, 1954.

BILIARY CIRRHOSIS-CYSTIC FIBROSIS SYNDROME

Synonymy

S. of focal biliary cirrhosis. S. of multilobar biliary cirrhosis with stones.

Syndromography

Clinic

Incidence: 4-22% of patients with cystic fibrosis present this anatomoclinical form. These cases of cystic fibrosis evolve in three stages:

Stage I. Biliary cirrhosis with asymptomatic stones.

Stage II. Multinodular biliary cirrhosis with hard liver nodules on palpation.

Stage IIIa. Hepatosplenomegaly with portal hypertension.

Stage IIIb. Hypersplenism, anemia, gastrointestinal bleeding, ascites.

In addition, lesions occur in other organs typical of cystic fibrosis (pancreas, lung, etc.).

Paraclinical examinations

Stage I. Negative liver tests. Sweat electrolyte tests, positive. Radiology of cystic fibrosis (see cystic fibrosis).

Stage I. The same results are maintained as in the stage I tests.

Stage II. Decreased albumin, increased alpha globulin fraction, anemia, thrombocytopenia, hyperbilirubinemia. Increased portal pressure.

For the three stages:

Radiology. Esophageal varices.

Liver biopsy. Bile ducts obstructed by eosinophilic amorphous material, surrounded by marked fibrosis.

Proliferation of bile ducts. Marked distortion of hepatic lobules, true cirrhotic areas, alternating with totally normal areas.

Syndromogenesis and etiology

Primary mechanical obstruction of the bile ducts due to secretion and thick bile as an expression of abnormal secretion products of the exocrine glands, typical of cystic fibrosis.

Ninety-six percent of patients with cystic fibrosis and evidence of liver disease had biliary tract obstruction with narrowing of the common bile duct.

Its etiology is related to cystic fibrosis, it is genetic and the most frequent mutation is deltaF 508 of autosomal recessive inheritance (see cystic fibrosis).

Literature

Gaskin, K.J. et al.: Liver disease and common-bili-duct stenosis in cystic fibrosis." N. Eng. J. Med., 318:340-346, 1988.

CYSTIC SYNDROME

Synonymy

S. of the stasis vesicle. S. of dyskinesia of the neck of the gallbladder. S. cystic-cholecystic. S. of organic syphopathy of the gallbladder. S. of the cystic duct. S. infundibulocystic.

Syndromography

Clínica

Prevalence in multiparous women. Stabbing pain in the gallbladder area, sometimes radiating to the back after a fatty meal.

Physical examination. Pain on palpation in the gallbladder region. Subictero sometimes.

Paraclinical examinations

Cholecystography and biliary drainage. With or without cholecystokinin, they allow us to see the change in appearance of the vesicle during contraction. They often reproduce the pain.

Syndromogenesis and etiology

Partial mechanical obstruction of the cystic, non-lithiatic.

Strong contractions of the gallbladder trying to overcome the obstacle to the free flow of bile.

Bands that compress the cystic, infundibulo-cystic angulation due to vesicular adhesions, and congenital or acquired cystic stenosis have been reported.

Literature

Camishion, R.C. and F. Goldstein: "Partial noncalculous cystic duct obstruction (cystic duct syndrome)." Surg. Clin. N. Amer.. 47:1107-1114, 1967.

Schieden, V.: "Uber die Stauungs-gallenblase." Zentralbl. Chir., 41:1257, 1920.

PRIMARY SCLEROSING CHOLANGIITIS SYNDROME

Syndromography

Clínica

This syndrome is more common in men (60-70%) and begins before the age of 45. It is characterized by signs of chronic or intermittent biliary obstruction: jaundice, pruritus, pain in the right upper quadrant and/or acute cholangiitis with fever. In advanced stages, biliary obstruction becomes permanent and leads to secondary biliary cirrhosis, liver failure or portal hypertension with or without bleeding varices.

Physical examination. Hepatomegaly, splenomegaly and of course jaundice can be detected in the acute phases of the syndrome.

It is almost always associated with chronic inflammatory bowel disease such as ulcerative colitis: or with multifocal fibrosclerosis syndromes: retroperitoneal, mediastinal, periurethral fibrosis, Riedel's stroma, or pseudotumor of the orbit.

Paraclinical examinations

Liver function tests. They are those of intense cholestasis: high hyperbilirubinemia at the expense of conjugated bilirubin. Elevated alkaline phosphatase, an important fact that can be found even in the anicteric phases. Elevated total cholesterol and lipids. High plasma levels of IgM exist in 50% of patients.

Endoscopic retrograde cholangiography. It is the method of choice and shows rosary-shaped narrowings of the main bile ducts, either in some or all.

Laparoscopic cholecystocholangiography. Shows the same more alterations.

Syndromogenesis and etiology

The syndrome is due to fibrosis and obstruction of the extrahepaic and intrahepatic bile ducts at any level including the sphincter of Oddi. Immunological factors have been pointed out as the cause of the syndrome.

The differential diagnosis must be considered with other lesions that affect the bile duct: tumors, the obstruction is slow and progressive, it may not be accompanied by pain or fever; If it is lithiasis or parasites, the symptoms are intermittent and reproduce the described picture.

The etiology is unknown.

Literature

Wiesner R.H.; N.F. La Russo: "Clinic pathologic features of the syndrome of primary sclerosing cholangitis." Gastroenterology, 79: 200-206, 1980.

Chapman, R.W.; B.A. Arborgh; J.M. Rhodes; et al.: "Primary sclerosing cholangitis: A review of its clinical features, cholangiography and hepatic histology." Gut, 21:870-877, 1980.

PANCREATIC CHOLERA SYNDROME

Synonymy

Diarrhea-producing islet cell tumor S.

Vipoma. S. of watery diarrhea-hypokalemia. S. de Verner-Morrison.

Syndromography

The acronym WDHA stands for watery diarrhea hypokalemia achlorhydria. In Spanish we can consider it: DLHA, that is, liquid diarrhea, hypokalemia, achlorhydria.

Clinic

It occurs in the second to sixth decades, predominantly in the fifth. There is diarrhea of varying intensity and duration, generally progressive. In the acute phases, diarrhea is watery and is accompanied by abdominal pain, nausea and vomiting. Secretory diarrhea is often profuse and often causes shock and renal failure. It is frequently nocturnal and persists in fasting. During the non-acute stages, there are thick stools and weight loss.

In general, there is great weakness that progresses to produce paralysis and stupor. It is sometimes accompanied by hot flashes. The duration of symptoms can be from months to years and curiously remissions occur in women during pregnancy.

Physical examination. Weight loss. Hypotonia and signs of dehydration.

Paraclinical examinations

Blood. Hypokalemia in all cases. Frequently hypercalcemia (76%) and hyperglycemia.

Coproculture. Negative.

Gastroacidogram. Basal anachlorhydria in two-thirds of cases with histamine-responsive hypochlorhydria.

Radiology. Absence of gastroduodenal ulcer or gastric fold hypertrophy.

Ultrasound and computerized axial tomography. In many cases, they allow the tumor to be located in the pancreas, which makes its removal possible and also determines the presence or absence of liver metastases.

Syndromogenesis and etiology

At present, DLHA syndrome tends to be considered as caused by a VIP-producing pancreatic endocrine tumor, that is, a vipoma, producing vasoactive intestinal peptide, in order to try to explain most of the symptoms by a single etiology. It is suggested that it originates from the delta cells of the pancreatic islets. 50% of cases are malignant. It has also been suggested that it is an exaggerated secretion of pancreatic peptide, gastric polypeptide, and prostaglandin E.

The mechanism of diarrhea is unclear, it may be due to excessive secretion of enteric juice. Hypokalemia may be due to loss of potassium with diarrhea or lack of absorption of this electrolyte.

Literature

Matsumoto, K.K.; J.B. Peter; R.G. Schultze, et al.: “Watery diarrhea and hypokalemia associated with pancreatic islet cell adenoma.” Gastroenterology, 50:231-242, 1966.

Moldawer, M.P. and G.L.Nardi: "Adenomas of the parathyroids and pancreatic islets with tumor of the pituitary: a syndrome with familial incidence." Amer. J. Med, Sci.. 228:190-206, 1954.

Verner, J.V. and A.B. Morrison: “Islet cell tumor and a syndrome of refractory watery diarrhea and hypokalemia." Amer. J. Med., 25:374-380, 1958.

BENIGN RECURRENT CHOLESTASIS SYNDROME

Synonymy

S. of benign intrahepatic cholestasis. S. of intrahepatic obstructive jaundice. S. of Summerskill- Walshe.

Syndromography

Clinic

This syndrome has a certain predominance in men. Its first episode appears before the age of 20. Jaundice is preceded by a sometimes long prodromal period, up to 1-2 months.

It is characterized by anorexia, asthenia, general malaise, sometimes nausea and/or vomiting with or without abdominal pain. Sometimes it is preceded by symptoms of urinary or respiratory infection.

There is intense jaundice accompanied by marked pruritus.

Acholia and coluria.

Physical examination. Discrete hepatomegaly. No splenomegaly.

Traces of scratching. Jaundice that can be melanistic when the episode lasts several months. Pain in the right upper quadrant (50%).

Paraclinical examinations

They correspond to cholestasis.

Blood. Hyperbilirubinemia at the expense of the conjugated fraction. Elevated alkaline phosphatase, moderately increased cholesterol, greatly increased bile acids. Modification (variable) of transaminases. Viral hepatitis markers are normal.

Laparoscopy. Smooth liver of intense green color. Normal or flaccid gallbladder. No splenomegaly. No alterations of the abdominal cavity.

Cholangiography. It demonstrates normality of the extrahepatic and intrahepatic bile ducts.

Liver biopsy. Obvious signs of cholestasis: bile stasis in the canaliculi. Presence of round cells in the portal spaces. Normality of hepatocytes and lobules.

Electron microscopy. During jaundice episodes:

Numerous vesicles with low-density material are observed in many hepatocytes. This disappears in referrals.

Syndromogenesis and etiology

This syndrome behaves like a pure cholestasis with no history, due to a defect in the excretion of conjugated bilirubin by the liver cell towards the canaliculi.

It has been concluded that the defect is not mediated by a circulating cholestatic agent, but rather that this syndrome is secondary to an intrinsic abnormality in the excretion of bile salts from the hepatocyte to the canaliculus.

From an etiological point of view, it appears to be caused by a genetic defect, which is transmitted in an autosomal recessive manner.

Literature

Aagenaes, O.; C.B. Van Der Hagen and S. Refsum: “Hereditary recurrent intrahepatic cholestasis from birth'. Arch. Dis. Child. 43:646-657, 1968.

Llanio, R.; A. Sotto; 0. Nodarse and M. Quintero: "Recurrent benign cholestasis. Review of the entity and report of the first case in our country." Rev. Cub. Med., 12(3):259-289, May0-June 1973.

Minuk, G.Y. and E.A. Shaffer: "Benign recurrent intrahepatic cholestasis evidence for an intrinsic abnormality in hepatocyte secretion." Gastroenterology, 93:1187-1193. 1987.

Summerskill W.H. and J.M. Walshe: "Bening recurrent intrahepatic obstructive jaundice." Lancet, 1:686-690, 1959.

IDIOPATHIC ULCERATIVE COLITIS SYNDROME

Synonymy

S. of severe colitis. S. of ulcerohemorrhagic colitis. S. of chronic suppurative colitis. S. of thromboulcerative colitis. S. of non-specific ulcerative colitis.

Syndromography

Clinic

There is the same incidence in both sexes. It begins at any age but is more common between 20-40 years, generally abruptly: diarrhea day and night, soft, bloody. Tenesmus and abdominal pain of varying degrees that is relieved by defecation. Anxiety preceding attacks.

Weight loss, sometimes fever. There are frequently manifestations of rheumatoid arthritis and less frequently erythema multiforme. Three clinical patterns can be differentiated:

1. Relapse remitter type.
   a) No fever, limited to attacks lasting 1-3 months.
   b) Severe, with fever, hemorrhage and toxemia.
2. Continuous, chronic type. Attacks lasting 6 months or more of varying intensity.
3. Acute fulminating type (5%). Fever, hemorrhages, perforation or obstruction.

Physical examination. During attacks: paleness, weakness, weight loss, slight pain throughout the colon, pain on rectal examination, signs of nutritional deficiency.

Paraclinical examinations

Blood. Microcytic anemia. Hypoproteinemia. Electrolyte alterations.

Radiology. Colon retracted, smooth, tubular. Loss of haustros.

Rectosigmoidoscopy and colonoscopy. It is the most faithful procedure: it reveals lesions that can range from hyperemic granular mucosa to ulcerations, bands and/or pseudopolyps; The lesions can bleed at the slightest contact with the equipment, since the mucosa is very friable. These alterations are dependent on the evolutionary state of the process.

Syndromogenesis and etiology

Anatomical alterations are the causes of the clinical symptoms and evolution, and the prognosis depends on them.

The etiology is unknown, although immunological, bacterial, genetic predisposition, emotional factors, etc. have been pointed out.

Literature

Roth, J.LA.: Ulcerative Colitis. In Bockus, H.L.: Gastroenterology. ed. 3, Philadelphia, Saunders, 1980.
Wilks, W. and W. Moxon: Lectures on Pathological Anatomy. London, Churchill, 1875.

IRRITABLE COLON SYNDROME

Synonymy

S. of colonic neurosis. S. of spastic colon. S. of mucosal colitis. S. of neuromuscular irritability of the colon. (See irritable bowel syndrome.)

Syndromography

Clinic

Triggering factors: psychogenic, cancerophobia, emotional tension, fatigue, menstrual period, tobacco, diet, intestinal irritants. It is characterized by constipation that alternates with diarrheal attacks (25%). Epigastric or colon pain. Associated symptoms: aerophagia, anxiety, insomnia, nervousness and fatigue.
Vasomotor symptoms: hot flashes, sweating, cardiac arrhythmia, headache, frequency with dysuria, dysmenorrhea, anovulvar pruritus.

Physical examination. During the attack: pain on palpation of the colon with spasticity especially in the descending and transverse portion.

Children. Start between 6 months and 2 years. Outbreak diarrhea with periods of constipation: consisting of one hard, abundant stool, followed by three to four small, loose, lienteric stools with mucus. Nutrition and appetite preserved. Some sporadic abdominal discomfort.

Paraclinical examinations

Stool. Macroscopic alterations depend on the stage. Absence of parasites or infections.

Sigmoidocolonoscopy. Spastic colon, with slight hyperemia of the mucosa. No injuries.

Radiology. Food with barium: increased traffic speed. In the colon by enema there are no important or characteristic signs.

Syndromogenesis and etiology

Emotional factors. It can be a manifestation of various endocrinopathies.

Literature

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Howship, J.: "Practical remarks on the discrimination and successful treatment of spasmodic stricture in the colon considered as an occasional cause of habitual confinement of the bowel." 1830.
Quoted by Ryle, J.A.: "Chronic spasmodic affections of the colon. Lancet, 2:1115-1119,1928.
Manousos, O.N.; S.C. Truelove and K. Lumsden: "Transit times of food in patients with irritable diverticulosis colon syndrome and normal subjects." Br. Med. J., 3: 760-761, 1967.
Ryle, J.A.: "Chronic spasmodic affections of the colon." Lancet, 2:1115-11 19, 1928.

COMFORT-STEINBERG SYNDROME

Synonymy

S. of relapsing chronic pancreatitis. S. of hereditary pancreatitis,

Clínica

It begins in childhood or adolescence, 4-15 years, in acute or chronic form. Recurrent episodes of intense abdominal pain radiating to the back. Nausea or vomiting.

Years later, the symptoms of diabetes mellitus develop. In other family members, diabetes may exist without these previous symptoms.

Physical examination. During attacks the symptoms are similar to those of acute pancreatitis.

Paraclinical examinations

Radiology. Pancreatic calcifications.

Blood. Increased amylase and lipase during crises.

Determination of pancreatic enzymes.

Duodenal intubation with stimulation.

Syndromogenesis and etiology

In vitro studies suggest that a specific protein "inhibits the nucleation of calcium carbonate and decreases the rate of crystal growth, which suggests that this protein acts as an inhibitor of the spontaneous formation of calcium carbonate in supersaturated pancreatic juice, a similar function is observed in human saliva. This protein was absent in a case of calcifying pancreatitis, indicating that its absence could explain the syndromogenesis of this syndrome.

The etiology is monogenic and is transmitted in an autosomal dominant manner; the mutated allele could be related to the absence of this protein.

Literature

Comfort, M.W.; E.E. Gambill and A.H. Baggenstoss: "Chronic relapsing pancreatitis: a study of 29 cases without associated disease of biliary or gastrointestinal tract." Gatroenterology, 6:239-285, 1946.

Comfort, M.W. and A.G. Steinberg: *Pedigree of family with hereditary chronic relapsing pancreatitis." Gastroenterology, 21:54-63, 1952.

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Sarles, H.; A. de Caro; L. Multigner and E. Martin: "Giant pancreatic stones in women due to abscense of the stone protein?* (Letter), Lancet, I:714-715, 1982.

NIGHT EATING SYNDROME

Synonymy

Morning hyperphagia-insomnia-anorexia syndrome.

Syndromography

Clinic

This syndrome predominates in women. They are generally obese patients or those with weight disorders. They suffer from insomnia and hyperphagia during the night with anorexia in the morning.

Syndromogenesis and etiology

It seems to be a response to stress in patients with emotional disorders.

The etiology is unknown.

Literature

Stunkard, A.J.; W.J. Grace and H.G. Wolff: "The nighteating syndrome. A pattern of food intake among certain obese patients." Am. J. Med., 19: 78-86, 1955.

COURVOISIER-TERRIER SYNDROME

Synonymy

S. of obstruction of the toilet ampulla.

Syndromography

Clinic

It is characterized by persistent, progressive jaundice that sets in slowly. This jaundice is apyretic and painless, unlike common bile duct syndromes, especially gallstones, in which these symptoms and chills are present and can disappear in a few days. This syndromic picture is completed by the presence of intense coluria and hypocolic and acholic stools, like putty or mastic. The itching is constant and is a cause of insomnia. Anorexia, asthenia and weight loss complete the syndromic picture.

Physical examination. Jaundice is observed in the conjunctivae, mucous membranes and skin, which acquires a melanic tinge.

Hepatomegaly and especially vesiculomegaly are palpable. Scratch marks on the skin are frequent.

Paraclinical examinations

Blood. High hyperbilirubinemia with predominance of unconjugated bilirubin. Elevated alkaline phosphatase. Normal or slightly increased transaminases. High cholesterol. In summary, the alterations are associated with marked cholestasis. Anemia and increased erythrocyte sedimentation rate.

Urine, intense coluria.

Syndromogenesis and etiology

This syndrome was described in relation to obstruction of the ampulla of Vater, but neoplasms of the head of the pancreas or duodenum close to the ampulla and even primary ones of the common bile duct can reproduce all the elements of the syndrome. Bile retention explains all the symptoms.

CROHN SYNDROME

Synonymy

S. of regional enteritis. S. of regional ileitis. S. of granulomatous ileocolitis.

Syndromography

Clinic

This syndrome is more common in Jewish descendants. It begins at any age and on average at 25 years old.

Symptoms vary with the location of the lesions and their intensity. Most commonly, it begins insidiously with borborygmi, flatulence, cramp-like abdominal pain, and mild diarrhea. Sometimes fever.

This condition progresses with the symptoms intensifying:

1. Pain (85% of patients) of colicky type with or without diarrhea that is relieved by defecation. It is sometimes confused with acute appendicitis. It is located in the periumbilical region or in the right iliac fossa. When the disease progresses, the pain becomes fixed and is located at the site of the injury.
2. Diarrhea (90%) soft or liquid.
3. Fever (33%) without chills.
4. Perianal and/or perirectal fistulas.
5. Symptoms of intestinal obstruction with or without perforation, especially when the lesion is located in the stomach or duodenum, accompanied by nausea, vomiting and pain reminiscent of an ulcer.
6. Mane (4%).
7. Systemic manifestations: fever, weight loss, polyarthritis (5%), erythema nodosum (rare), pyoderma gangrenous.

Physical examination. Abdominal distension, painful palpation in the affected region. Mass that can be palpated in the lower right drant.

Paraclinical examinations

Blood. Moderate anemia, leukocytosis, hypoproteinemia, hypoprothrombinemia, hypocalcemia, electrolyte alterations.

Radiology. Dilations with narrowing of the lumen and thinning of the villi. Longitudinal ulcers.

In the advanced periods of the disease, the appearance at the lesion location is a rigid tube with loss of the mucosal pattern and a bird's beak shape. Apparently healthy areas intertwined with injuries in some cases. The "stoned" appearance is characteristic and sometimes fistulous tracts are observed.

Colonoscopy and biopsy. It is the most accurate method to define the diagnosis, the evolutionary period and the surveillance of a possible superimposed cancer when the lesions are located in the colon. In those cases in which the lesion is located in the terminal portion of the small intestine (most common), this method can be useful because in a good number of examinations it is possible to reach that level and take a biopsy.

Syndromogenesis and etiology

It is proposed to include Crohn's syndrome in the group of autoimmune diseases.

Cases have been reported in families in 10% of patients with regional enteritis and one or more close relatives have been found to have granulomatous disease of the large intestine.

The family pattern does not suggest a simple Mendelian inheritance, although it does suggest genetic involvement that has not yet been specified.

The etiology is unknown.

Literature

Morgagni (1769) quoted by Goldstein, F.: Regional Enteritis. In Bockus, H.L.: Gastroenterology. Philadelphia, Saunders, 1984.

Orholm, M., et al: "Familial occurrence of inflammatory bowel disease." N. Engl. J. Med. 324:84-88, 1991.

CRONKHITE SYNDROME -CANADA

Synonymy

S. of polyposis-pigmentation-alopecia-onychotrophy. S. of diffuse gastrointestinal polyposis with ectodermal alterations.

Syndromography

Clínic

This syndrome is prevalent in women. It is characterized by: diffuse brown pigmentation of the skin, more intense in the folds of the body; generalized alopecia; onychodystrophy manifested by the formation of a new nail beneath the superficial portion of the old abnormal nail.

These symptoms may precede or follow digestive symptoms: nausea, vomiting, chronic diarrhea, diffuse inflammatory gastrointestinal polyposis, preferably in the small intestine, but also in the colon and stomach. Protein-losing enteropathy, malabsorption, anemia and edema.

Paraclinical examinations

Radiology of the gastrointestinal tract. Polyposis in the affected segments. Confluence of the intestinal folds with or without defect due to polyps. Segmentation of barium in intestine.

Endoscopy with biopsy. Visualization of polyps.

They are generally adenomatous.

Syndromogenesis and etiology

No familial hereditary factors have been demonstrated.

The extension of the polyps can cause malabsorption and protein loss syndrome, which improve with removal of the polyps. It has also been suggested that a disaccharidase deficiency and bacterial overgrowth are the cause of diarrhea and malabsorption.

The etiology is unknown.

Literature

Cronkhite, L.W. and W.J. Canada: "Generalized gastrointestinal polyposis: Unusual syndrome of polyposis, pigmentation, alopecia and onychotrophia." N. Engl. J. Med., 252:1011, 195.

Johnson, G.K. et al.: "Cronkhite-Canada syndrome: Gastrointestinal pathophysiology and morphology." Gastroenterology, 63:140, 1972.

CRUVEILHIER-BAUMGARTEN SYNDROME

Synonymy

Baumgarten cirrhosis. S. de Pegot-Cruveilhier-Baumgarten.

Syndromography

Clinic

Digestive disorders. Hematemesis.

Physical examination. Abdominal distention, marked dilation of the veins from the navel in the shape of a medusa head, abdominothoracic collateral circulation, venous murmur and thrill (shivering) in the periumbilical region. Atrophic liver.

Chronic congestive splenomegaly. Ascites rarely occurs.

Paraclinical examinations

Radiology. Esophageal veins, portal hypertension with obstruction of the portal within the liver. Umbilical vein patency.

Laparoscopy. It demonstrates atrophic cirrhosis, splenomegaly and a marked increase in venous collaterals due to marked portal hypertension. It allows us to see in its entirety the persistence and dilation of the umbilical vein of the fetus from the liver to the inner wall of the navel. Sometimes there is what we have called internal Cruveilhier-Baumgarten, since these radial dilations are observed like those described, but only in the peritoneum of the internal umbilical wall.

Syndromogenesis and etiology

This syndrome always occurs in cirosis with splenomegaly and portal hypertension, but with persistent permeability of the umbilical vein. This is the fundamental element that explains this syndrome.

Literature

Baumgarten, P. von: Über vollständiges; Ofenbleiben der vena umbilicalis; Zugleich ein Beitrag zur Frage des Morbus Banti in: Baumgartens Arbeiten". Leipzig, 6:93, 1907.

Cruveilhier, J.: Anatomie Pathologique du Corps Humain. Vol I (Paris: Chez J. B. Baillière, 1829-1835), p. 16.

CHILAIDITI SYNDROME

Synonymy

S. of hepatodiaphragmatic interposition. S. of interposi-subphrenic tion.

Syndromography

Clínica

Adults are asymptomatic or have few symptoms.

Children present: frequent abdominal pain associated with vomiting that appears at the end of the day when the distension is greater and is relieved by lying down. Less frequently constipation and gas noise.

Physical examination. Abdominal distension, absence of hepatic dullness, pseudohepatomegaly due to descent of the liver.

Paraclinical examinations

Radiology. Partial or total interposition of the colon between the diaphragm and the liver.

Syndromogenesis and etiology

The interposition of the colon between the diaphragm and the liver can cause intestinal distention, paralysis of the diaphragm and

abnormalities in the liver ligaments. In children, aerophagia and/or abdominal distention are the main mechanisms.

The etiology is multiple and may be due to: diaphragmatic causes (congenital anomalies, eventrations, phrenic paralysis); hepatic causes (absence of the right lobe, hepatic ptosis, ligament abnormalities); intestinal causes (dolichocolon or megacolon, long intestinal meso).

Literature

Borhammanesh: "Hepatodiaphragmatic interposition of the intestine. Chilaiditi'"s syndrome". Clin. Radiol. (Edinb.), 27:113- -116, 1976.

Chilaiditi, D.: "Zur Frage der Hepatoptose in Anscluss an Falle Von temporärer partieller Leberverlagerung." Fortschr. Röntgenstr., 16:173-208, 1910.

CHIRAY SYNDROME

Synonymy

S. of atony of the gallbladder. S. of biliary hypotonia. S. of biliary dyskinesia. S. of lazy vesicle.

Syndromography

Clínica

Anorexia, nausea, dyspepsia with intolerance to fats, pain attacks in the right upper quadrant radiating to the back, headache and nervous depression.

Physical examination. Pain on palpation of the right hypochondrium.

Paraclinical examinations

Cholecystography. It shows a dilated gallbladder with delayed emptying.

Biliary drainage. Delay in the emptying of bile B that has a very dark brown or black color.

Syndromogenesis and etiology

It is due to a motor disorder of the gallbladder that leads to gallbladder atony with stasis, distension and pain. Other disorders that produce distention and hyperpressure of the gallbladder, such as spasm of the sphincter of Oddi (functional or organic), cause the same symptoms.

Literature

Chiray, M.: “Etat current de le cholecystoatonic.” In: Albot e Poilleux: Les voies biliaires. Paris, Masson, 1953.

LACTASE DEFICIENCY SYNDROME

Syndromography

Clínica

When the patient ingests milk or lactose, the following occur: borborygmi, flatulence, colic. If the amount ingested is greater, diarrhea occurs. This causes patients to avoid milk and its derivatives. It is much more common in blacks and some races such as Bantus and Orientals than in whites.

The symptoms are not due to a food allergy to milk proteins, but to the lactose contained in it. The ingestion of lactose alone reproduces them.

It is curious that although it is of genetic origin, the symptoms sometimes appear during puberty.

Paraclinical examinations

Lactose tolerance test. False results (20%) due to retention in the stomach must be taken into account. Measurement of breath hydrogen after ingestion of 50 g of lactose is more specific and sensitive. The explanation is that bacteria in the colon release hydrogen from unabsorbed lactose and therefore increases elimination through the breath.

Syndromogenesis and etiology

This syndrome has a genetic etiology with autosomal recessive inheritance. The term lactase restriction has been used to explain homozygous recessive cases, in which people with this genotype have a low lactose digestion capacity. The lactase of the ciliated epithelium of the small intestine also has phlorizin-hydrolase activity; This enzymatic activity is the one that is most frequently affected in this syndrome.

It has been proposed that in adults with low lactase activity, it begins to decrease approximately between 3-5 years of age, which suggests that the lactase expression pattern is regulated at the level of gene transcription.

Literature

Bond, J.H. and M.D. Levit: "Use of breath hydrogen in the study of carbohydrate absorption." Am. J. Dig. Dis. 22:379, 1977.

Montgomery, R.K., et al,: "Lactose intolerance and the genetic regulation of intestinal lactase-phorizyn hydrolase." FASEB J. 5:2824-2832, 1991.

DUODENAL STUMMP TEAR SYNDROME

Syndromography

Clinic

3-6 days after gastric resection, intense, constant pain, without irradiation, occurs in the upper right part of the abdomen, which remains localized at first. If it is not diagnosed, symptoms of peritonitis occur that slowly spread to the rest of the abdomen. Sometimes it progresses to the formation of an abscess or fistula.

Physical examination. Pain and contracture of the abdominal wall that intensifies. Tachycardia and fever.

Syndromogenesis and etiology

It is due to a dehiscence of the suture of the duodenal stump, which if not diagnosed early leads to generalized peritonitis with serious consequences.

Bibliography

Larsen, B.B. and E. C. Foreman: "Syndrome of leaking duodenal stump." Arch. Surg., 63:480-485, 1951.

DIEULAFOY SYNDROME

Synonymy

S. of simple ulcus. S. of Dieulafoy's acute solitary gastric erosion.

Syndromography

Clinic

The syndrome occurs especially in young people without a history of gastric symptoms. Without prodromes (only occasionally there is nausea or a sensation of epigastric heat). Massive hematemesis begins unexpectedly without food remains in the vomited material.

The patient is in danger of death in a few hours if the diagnosis is not specified and appropriate therapy is not installed.

Paraclinical examinations

Gastrochymogram. Normoacidity or hypoacidity.

Blood. Secondary hypochromic anemia proportional to hemorrhage. Red blood cells can reach two million and hemoglobin 4-6 mg per 100 dL. Hematocrit 10-15%.

Gastroscopy. Small ulcer with pulsating bleeding arteriole in most cases. It is the fastest and safest exam.

Syndromogenesis and etiology

It is due to an ulceration that crosses the muscularis muCOSae and erodes a small submucosal arteriole; Sometimes it is due to an angiomatous anomaly of the gastric wall or an aneurysmal vessel of the submucosa. The location is generally in the upper third of the stomach.

Literature

Dieulafoy, G.: Clin. Med. Paris 263, 1897-1898.

Dieulafoy, G.: Ulcération gastriques'". Sem. Méd. Paris., 263, 1900.

Weissenhofer, A.: "Die akutte Blutung bei Ulcus Dieulafoy.
Langenbecks". Arch. Chir., 341:135-140, 1976.

DILATION SYNDROME

SEGMENTARY COLON

Syndromography

Clinic

Constipation from birth, accompanied by dilation of part or the entire colon.

Paraclinical examinations

Radiology. The dilation of the affected segment of the colon is fusiform and indistinguishable from Hirschsprung's disease.

Biopsy. Uniform muscle hypertrophy in the dilated segment. Normal nerves and ganglion cells, which allows this syndrome to be distinguished from Hirschsprung's disease.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Ehrenpreis, T.: "Pseudo-Hirschsprung's disease." Arch. Dis. Child., 40:177-179, 1965.

Swenson, O. and F. Rathauser: *Segmental dilation of the colon". Amer. J. Surg., 97:734-738, 1959.

ESOPHAGEAL DYSYNERGY SYNDROME

Synonymy

S. of achalasia. S. of cardiospasm. S. of esophageal dystonia.
S. of aperistalsis of the esophagus. S. of functional hiatal stenosis.

Syndromography

Clinic

This syndrome begins at any age, although it is more common between the fourth and sixth decades. It has the same incidence in both sexes. It begins abruptly, sometimes preceded by minor symptoms: salivation for years, a sensation of obstruction at the level of the xiphoid process. Pain or discomfort when eating food. The episodes are intermittent for a time, they improve by eating slowly and drinking fluid frequently during food intake. When the disease is more advanced, intense substernal pain occurs after meals, especially in some variants of the syndrome. Regurgitation relieves symptoms. This regurgitation is without nausea or a foul odor, but is rich in mucus.

Physical examination. Determination of swallowing time with the stethoscope placed on the xiphoid. In some cases there is dilation of the neck veins and cyanosis after eating, which are relieved by regurgitation. Pallor.

Paraclinical examinations

Radiology. Esophagus dilated and ending in a pencil point.
Endoscopy. Check for dilation, food debris and mucus. Mucosal hyperemia. Curiously, the endoscope can pass the narrowing with relative ease in most patients.

Motility. Altered and must be complemented with the methacholine test, which demonstrates great sensitivity.

Syndromogenesis and etiology

There is a neuromuscular dysfunction of the esophagus, especially a defect in cholinergic innervation. There is also a psychogenic component in most cases.

The etiology is unknown.

INTESTINAL DYSPEPSIA SYNDROME TO CARBOHYDRATES

See irritable bowel syndrome.

Syndromography

Clínica

This syndrome predominates in middle-aged women, who have a tendency to suffer from depressive symptoms: anxiety, nervousness. Characteristic intermittent attacks after emotional stress. These can last days or years. Abdominal pain usually after meals with nighttime discomfort.

Feeling of fullness and abdominal distention with a lot of gas, intolerance to certain foods (carbohydrates, vegetables, milk), constipation or diarrhea, fatigue.

Physical examination. Weight loss in some patients. Choroidoretinitis (25%), hypotension (30%), tachycardia.

Paraclinical examinations

Blood. Moderate anemia.

Stool. Rich in undigested carbohydrates; increased mucus and gas bubbles.

Sigmoidocolonoscopy. Normal.

Radiology. Colon dilation, spasms and transit accelerated.

Syndromogenesis and etiology

Alteration of intestinal flora, enzyme deficiency, intestinal hypermotility, sensitivity to carbohydrates, aerophagia.

Literature

Bargen, J.A.: “Irritable bowel syndrome.” Writings & Reports from the Scott and White Clinic, 4:57-62, 1965-1966.

SMALL INTESTINE DIVERTICULOSIS-MACROCYTIC ANEMIA SYNDROME

Synonymy

S. of small intestine diverticulosis-macrocytic anemia-malabsorption. S. of jejunal diverticulosis- -megaloblastic anemia.

Syndromography

Clinic

This syndrome includes all types of megaloblastic anemia that present with free hydrochloric acid and intrinsic factor in the stomach. We refer to: Lichtheim syndrome, diverticulosis of the jejunum (occasionally also diverticulosis of the duodenum, esophagus, stomach, colon and urinary bladder) associated or not with malabsorption syndrome (steatorrhea).

Paraclinical examinations

Blood. Megaloblastic anemia.

Bone marrow. Megaloblastic changes.

Radiology. Multiple diverticula of the intestine that vary in size from millimeters to several centimeters.

Motility. Functional motor alteration of the small intestine.

Stool. Steatorrhea.

Syndromogenesis and etiology

The etiology is unknown. Possibly congenital.

Literature

Badenoch, J., et al.: "Massive diverticulosis of small intestine with steatorrhea and megaloblastic anemia." Q.J.Med., 24:321, 1955.

Irwin, G.A.: "Syndrome of jejunal diverticulosis and megaloblastic anemia." Am. J. Roentgenol., 94:366, 1965.

DUBIN-JOHNSON SYNDROME

Synonymy

Dubin-Sprinz disease. Icterus-hepatic pigmentation. S. of black liver-jaundice. S. de Sprinz-Nelson. S. of idiopathic chronic jaundice. S. of familial conjugated hyperbilirubinemia.

Syndromography

Clínica

Dubin-Johnson syndrome almost always begins in adulthood. According to some authors: in the first year of life, 12%; before the age of 15, 38%. However, a quarter of patients remain asymptomatic throughout their lives.

Intermittent episodes of jaundice are associated with mild pain in the right upper quadrant. Hypocolia and choluria, absence of pruritus, anorexia, nausea, vomiting, asthenia and bleeding tendency due to factor VII deficiency.

Physical examination. Liver of normal size or slightly enlarged.

Paraclinical examinations

Blood. Increased conjugated bilirubin. However, thanks to the newest and most precise method (alkaline methanolysis and high pressure chromatography) it has been shown that homozygous patients with this syndrome have significant concentrations of unconjugated bilirubin in the serum. Furthermore, serum contains more diconjugated than monoconjugated bilirubin, exactly the opposite of what happens in acquired diseases and Rotor syndrome.

This inversion of the proportion is considered characteristic and serves as a diagnosis in homozygous patients.

Alkaline phosphatase. Normal.

Bromosulfthalein excretion test. Altered with retention in the blood 2 h after injection. This is a product of the reflux of conjugated contrast material in the liver and indicates the defect in maximum hepatic excretory transport (TM). However, this does not happen with indocyanine green, since it is not conjugated in the liver.

Urine. Bilirubinuria.

Radiology. Oral or intravenous cholecystography does not stain the gallbladder.

Laparoscopy. It makes the diagnosis easy by showing an almost black liver in the presence of discrete jaundice.

Liver biopsy. It finds melanin-like pigment in liver cells, especially in centrilobular areas. Normal bile canaliculi. No round cell infiltration or fibrosis. There are no pigments in Küpffer cells.

Syndromogenesis and etiology

Functionally there is a defect in the biliary excretion of bilirubin, cholephilic materials and porphyrins.

The pigment has been shown to be free radical free in the absence of light, distinguishable from melanin or its related polymers.

Segregation analyzes have shown that it is transmitted by autosomal recessive inheritance with reduced penetrance.

Literature

Butt, H.R.; V.E. Anderson; W.T. Foulk, et al: "Studies of chronic idiopathic jaundice (Dubin-Johnson syndrome)." Gastroenterology 51: 619-630, 1966.

Dubin, I.N. and F.B. Johnson: "Chronic idiopathic jaundice with unidentified pigment in liver cells: new clinicopathologic entity with report of 12 cases." Medicine, 33:155-197, 1954.

Shami. M.. et al.: Dubin-Johnson Syndrome in Israel. "Cinical laboratory and genetic aspects of 101 cases. Quart J. Med., 39:549-567, 1970.

Sprinz, H. and R.S. Nelson: “Persistent non-hemolitic hyperbilirubinemia associated with lipochrome-like pigment in liver cells: report of 4 cases.” Ann. Int. Med., 41: 952-962, 1954.

Swartz, H.M.; K. Chen and J.A. Roth: "Further evidence that the pigment in Dubin-Johnson syndrome is not melanin. Pigment cell Res., 1:69-75, 1987.

DUDLEY-KLINGENSTEIN SYNDROME

Synonymy

S. of jejunal neoplasia.

Syndromography

Clinic

It presents as a combination of melena with abdominal pain, which simulates an ulcerative gastroduodenal lesion.

Physical examination. Pallor, tachycardia and abdominal distention.

Paraclinical examinations

Radiology. He is negative for gastroduodenal ulcer. Jejunography shows a filling defect more visible on double contrast radiography.

Gastroacidogram. Shows normal or low values of hydrochloric acid.

Blood. Posthemorrhagic anemia.

Syndromogenesis and etiology

This syndrome is due to a benign or malignant tumor of the jejunum.

Literature

Dudley, H.D.: Vascular tumors of the small intestine with symptoms simulating peptic ulcer." Surg. Clin. N. Amer., 14:1331-1337, 1934.

Klingenstein, P.: "Benign neoplasms of the small intestine complicated by severe hemorrhage: report of two cases operative intervention and recovery." J. Mount Sinai Hosp., 4:972-979, 1938.

Piccinni, L.: “Neoplasia of the jejunum with melena and pseudo-ulcer duodenal painful syndrome.” Clinica (Bologna), 15:33-39, 1954.

POST-GASTRECTOMY DEDUMPING SYNDROME

Synonymy

S. of Dumping. S. of postprandial distress. S. gastric remnant. Postgastrectomy pseudodeficiency syndrome. S. jejunal. S. postvagotomy.

Syndromography

There are two main types of dumping syndrome: early and late in relation to meals.

EARLY DUMPING SYNDROME

Known as true Dumping syndrome.

Clinic

This syndrome occurs immediately after eating or within 20-30 minutes. It is observed in 20-30% of gastrectomized patients.

Some patients have symptoms at every meal; others only once a day or with strong meals. It is characterized by a feeling of intense fullness, nausea and/or vomiting, borborygmi followed by diarrhea, vasomotor disorders such as: weakness, paleness, sweating (sweating, hyperhidrosis), tachycardia and sometimes syncope.

Paraclinical examinations

Blood. Hyperglycemia that coincides with the onset of symptoms. After slight decrease in potassium and phosphorus.

Electrocardiograph. Changes are detected.

Radiology. Symptoms can be reproduced by giving barium mixed with food or 50% glucose. Rapid emptying and dilution of the barium mixture within the intestine may be observed due to an increase in intraluminal fluid.

Syndromogenesis and etiology

In gastrectomized patients, there is a rapid filling of the jejunum with undigested food that constitutes concentrated and hyperosmotic solutions, which attracts a lot of fluid into the lumen and this decreases plasma volume.

Other mechanisms have been pointed out: stimulation of autonomic reflexes, release of intestinal hormones or both simultaneous phenomena that would occur in response to the rapid penetration of gastric contents into the duodenum and jejunum.

This syndrome is also observed after pyloroplasty, although less frequently.

LATE DEDUMPING SYNDROME

Clínic

In some gastrectomized patients, hypoglycemia occurs with all its symptoms (paleness, sweating, tremors, lipothymic state, etc.).

Syndromogenesis and etiology

Late Dumping is due to hypoglycemia that follows the peak increase in postprandial glycemia, where an exaggerated response of the pancreas with insulin release plays an important role.

Symptoms are relieved by ingestion of food or an appropriate diet.

Literature

Denecheau, D.: "Les suites medicales eloignees de la gastroenterostomie au cours de l'ulcere de l'estomac et de ses complications." Theses de Paris, 1907; Semaine med., 27: 316- -322, 1907.

Kiefer, E.D.: "Postgastrectomy syndromes." Am. J. Gastroenterol., 35:352, 1961.

HOOKWOOD DUODENITIS SYNDROME

Synonymy

S. de Griesinger.

Syndromography

Clinic

The symptoms are the same as those reported by a patient with a duodenal ulcer.

Paraclinical examinations

Gastroacidogram. The acidity is higher than that observed in patients with duodenal ulcer.

Radiology. Deformity of the duodenal bulb. Hyperperistalsis of the duodenum. No ulcer niche is observed.

Stool. The characteristic hookworm eggs are observed under a microscope.

Syndromogenesis and etiology

The syndrome is due to infestation by Ancylostoma duodenalis.

Literature

Yenikombishan, H. and W. Shehadi: "Duodenal ulcer syndrome caused by ankylostomiasis." Amer. J. Roentgen., 49:39-48, 1963.

JANBON ANTIBIOTICS ENTEROCOLITIS SYNDROME

Synonymy

S. anorectal. S. coliform. Pseudomembranous enterocolitis S. S. of postoperative enterocolitis.
Micrococcal enterotoxic S.

Syndromography

Clínic

This syndrome begins 2-30 days after the administration of broad-spectrum antibiotics in surgical interventions.

It is characterized by: watery diarrhea, similar to whey, which may be accompanied by mucus and blood, sometimes cramps and tenesmus. Foul-smelling vomiting, fever, oliguria and shock.
Marked electrolyte disorders with a tendency to metabolic acidosis.

Physical examination. Abdominal distension. Dehydration, hypotension, tachycardia.

Paraclinical examinations

Blood. Hemoconcentration, leukocytosis.

Stool. They contain remains of membranes, leukocytes and gram-positive micrococci.

Syndromogenesis and etiology

This syndrome is due to the enterotoxins of hemolytic staphylococcus aureus. This germ develops when the normal bacterial flora disappears due to the action of antibiotics.

Literature

Janbon, M., et al.: "Le syndrome choleriforme de la terramycine". Montpellier Med., 41:300-311, 1952.

CHOLERIC ENTEROPATHY SYNDROME

Syndromography

Clinic

This syndrome is observed in patients who suffer from ileal disease or who have undergone an ileal resection. The most frequent symptoms are: diarrhea with steatorrhea and progressive weight loss.

Paraclinical examinations

Stool. Determination of the amount of bile acids excreted in 24 hours. Determination of the fat absorption rate.

Blood. Cholesterol. Carotenes and vitamins D, E, K.

Syndromogenesis and etiology

The mechanism that produces this syndrome is the interruption of the absorption of bile salts due to a pathology of the ileum (site of bile salt reabsorption).

This is due to ileal resections, ileal diseases, ulcerative colitis with ileal seizure, Crohn's disease.
viral enteritis.

Literature

Hoffmann. A.F.: The syndrome of ileal disease and the broken enteropathic circulation: cholertheic enteropathy." Gasroenterology, 52: 752-757, 1967.

PROTEIN-LOSING ENTEROPATHY SYNDROME

Syndromography

Clinic

It has been calculated that under normal conditions 10-20% of protein turnover is due to loss through feces. In different processes this amount increases considerably, giving rise to a sustained loss of protein that leads to progressive malnutrition.

Syndromogenesis and etiology

The mechanisms indicated are:

1. Inflammation or ulceration of the mucosa that allows the passage of proteins as in regional enteritis, ulcerative colitis, intestinal tuberculosis, infectious enteritis, etc.
2. Alteration in the cellular structure of the mucosa as occurs in tropical sprue or not, where there is an abnormality of the villi and intestinal epithelium, as well as in celiac syndrome (allergy to gluten gliadin). The alterations may be in other mucous membranes such as those of the stomach; These are cases of Menetrier's disease, gastric carcinoma, etc.
3. Increased lymphatic pressure that occurs in granulomatous conditions or neoplasia of the abdominal lymph nodes, Whiple's disease, lymphoma, etc.
4. Dilated lymphatic vessels may rupture through the epithelium and discharge their contents into the lumen of the intestine as in idiopathic intestinal lymphangiectasia.

DEMOORE ABDOMINAL EPILEPSY SYNDROME

Synonymy

S. of abdominal epilepsy.

Syndromography

Clínic

This syndrome is common in children and adolescents. It is characterized by diffuse cramping abdominal pain, nausea and vomiting.

Paraclinical examinations

Electroencephalogram. Abnormalities are demonstrated.

Syndromogenesis and etiology

It is due to an injury to the cerebral cortex.

Literature

Moore, M.T.: Paroxysmal abdominal pain: form of focal symptomatic epilepsy." J.A. M.A., 124:561, 1944.

NON-TROPICAL SPRUE SYNDROME.

ADULT VARIETY

Syndromography

Clinic

This syndrome predominates in women (2:1). 25% of patients have a history of diarrhea in the first years of life, which disappears later. In some cases a malabsorption syndrome remains.

Symptoms generally begin between the third and sixth decades of life and are characterized by intermittent crises of exacerbation and remission, which are triggered by emotional stress. Also characteristic of this syndrome are: foul diarrhea, pasty stools (sometimes liquid), asthenia, weight loss, hemorrhage, manifestations of hypocalcemia, bone pain.

Physical examination. Arterial hypotension, ballooning and abdominal distention, with tympany on percussion, edema, glossitis and stomatitis, muscle hypotrophy. Occasionally clubbing fingers. Brown skin pigmentation that simulates Addison's syndrome.

Paraclinical examinations

Blood. Macrocytic anemia, hypoproteinemia, hypocalcemia, hypokalemia. hypolipemia, prolonged prothrombin

d-xylose test. Positive.

Urine. Determination of urinary indoles.

Stool. Steatorrhea.

Radiology. Barium segmentation and flocculation, thickening of the conniving valves of the jejunopleon (Monlage sign).

Jejunum biopsy. Lesions characteristic of edema, flattening of the villi, inflammatory infiltrate and alterations of intestinal cells.

Syndromogenesis and etiology

Deficiency of peptidases in the intestinal mucosa and therefore inability to hydrolyze diets with gluten.

Others assume that gluten may have some direct toxic effect on the mucosa in these cases.

It is transmitted by autosomal dominant inheritance of incomplete penetration.

The etiology is unknown.

Literature

Douglas, A.P. and C.C. Booth: "Jejunal mucosal digestion of gluten peptides in adult celiac disease." Lancet, 2: 491-492, 1968.

Gee, S.: "On the celiac affection." St. Barth. Hosp. Rep., 24:17- -20. 1988.

MacMahon, R.A.; M. LeMoine-Parker and M.C. McKinnon: "Zinc treatment in malabsorption" Med. J. Aust., 2:210-212, 1968.

TROPICAL SPRUE SYNDROME

Synonymy

S. of tropical diarrhea. S. of mouth ulcers from Ceylon (now Sri Lanka). Cochinchina Diarrhea S. S. of chronic intestinal indigestion. S. of fat intolerance. S. of intestinal infantilism.

Syndromography

Clinic

This syndrome is more common in tropical or subtropical regions, especially in white people. Insidious onset: explosive, foul-smelling, pale, frothy, high-fat diarrhea during the day and night.

Anorexia, nausea, vomiting, weight loss, paresthesia, pain in the mouth and anal region. The patient is indifferent to everything.

Physical examination. Paleness, cheilosis, stomatitis, thrush, glossitis with smooth and red tongue, teeth partially or totally absent. Manifestations of hemorrhage in the skin and subcutaneous or digestive tissues: moderate or severe. Severe exhaustion, premature aging, dry, brittle hair, arterial hypotension.

Abdomen: protruding, especially in standing position.

Liver of normal size, but smooth and soft to palpation.

Perform a neurological examination if there is associated Lichteim syndrome.

Paraclinical examinations

Blood. Macrocytic anemia, leukopenia, moderate thrombocytopenia, sometimes hypoprothrombinemia, hypocholesterolemia, hypoproteinemia, mild hyperbilirubinemia, normal serum iron and liver tests, vitamin A tolerance curve: flat.

d-xylose test. Flat curve.

Bone marrow. Megaloblastosis, giant metamyelocytes.

Gastrochymogram. Anacidity.

Stool. Increase in fatty acids.

Jejunum biopsy. Short, thin and molten villi given on many occasions. Mucosal infiltration with inflammatory cells and especially eosinophils.

Syndromogenesis and etiology

Alterations in the villi and intestinal cells explain the entire picture of this malabsorption syndrome.

The etiology is unknown

Literature

Hernández-Morales, F. and R.A. Sifre: Sprue. In Bockus, H.L.: Gastroenterology, v. I. Philadelphia, Saunders, 1980.

Hilary, W. and A. Severini: Observations on the Changes of the Air, and the Concomittant Epidemical Diseases in the Island of Barbadoes. ed. 2, London, Hawkes, Clarke, and Collins, 1766, p. 103.

Ketelaer, V.: Commentarius medicus, de aphthis nostratibus, seu Belgarum sprouw. Ludg. Bat. 1672.

ACUTE HEPATIC STEATOSIS SYNDROME OF PREGNANCY

Syndromography

Clinic

This syndrome occurs in young women in the third trimester of pregnancy. It is characterized by epigastric pain and/or

in the right upper quadrant, marked asthenia, nausea, vomiting. Episodes of: disorientation, confusion, stupor, coma and death.

Physical examination. Jaundice.

Paraclinical examinations

Blood. Increase in total bilirubin. Alterations in liver function tests.

Liver biopsy. Intense fatty infiltration of centrilobular location. Occasionally hepatocellular necrosis.

Syndromogenesis and etiology

The etiology is unknown. Different criteria are indicated:

1. Ingestion of medications during menstruation: tetracycline, especially intravenous, chlorothiazides.
2. Viral hepatitis acquired during pregnancy.
3. Deficiencies of lipotropic factors.
4. Hepatotoxins.

Literature

Czernobisky, B. and M.A. Bergnes: “Acute fatty metamorphosis of the liver in pregnancy with associated liver cell necrosis." Obstet. Gynecol., 26:792-798, 1965.

Peters, R.L.; HA. Edmondson and C.T, Kunelis: *Acute fatty metamorphosis of the liver in pregnancy". J.A.M.A., 180:767, 1962.

Peters, R.L.: H.A. Edmondson and C.T. Kunelis: "The clinical and pathological picture of acute fatty metamorphosis of the liver in pregnancy." Ann. Int. Med., 61:803-804, 1964.

FABER SYNDROME

Synonymy

S. of sideropenic anemia. S. of simple achlorhydric anemia. S. de Hayem-Faber. S. achylic0-chloroanemia-. S. of idiopathic hypochromic anemia.

Syndromography

Clinic

In general, it occurs in women between 30-40 years old, more frequently during convalescence from some diseases or after childbirth; rarely in men. It is characterized by general asthenia, menstrual disorders, weakness, drowsiness.

Physical examination. Pale skin with hyperpigmentation, perioral eczematoid lesions with rhagades; dry hair. Trophic alterations of the nails, koilonychia with painful formations on the tips of the fingers and toes. Precocious canicie. Glossitis and dysphagia.

In the circulatory system: tachycardia, anemic heart murmur.

Paraclinical examinations

Radiology: In the skeleton, a delay is observed in the nuclei of ossification and osteoporosis.

Blood. Hypochromic anemia, anisocytosis, microcytosis, poikilocytosis. In severe cases, leukopenia with relative lymphocytosis. Decreased serum iron. High iron storage capacity.

Medulogram. Erythropoietic hyperplasia with inhibition of maturation.

Gastrochymogram. Marked anachlorhydria.

Stool. Positive occult blood.

Syndromogenesis and etiology

This syndrome can be caused by different disorders that are associated with chronic blood loss: menstrual loss, fibromatous uterus, hemorrhoids, peptic ulcer, recurrent epistaxis, hereditary telangiectasia and others.

Due to the above, some authors have pointed out that this syndrome is obsolete; However, in the way originally described by Hayem and others, chronic hypochromic anemia is associated with aquilia, which maintains its individuality since this situation is not present in the other conditions mentioned.

This syndrome is caused by insufficient secretion of hydrochloric acid with increased intestinal transit, which determines insufficient absorption of iron.

Literature

Faber, K.: "Achylia gastrica mit Anämie" Med. Klin. 5:1310- -1312, 1909.

Faber, K.: "Om anaemistake Tilstande ved Achylia gastrica." Berl. Klin. Wschr., 50:958-962, 1913.

LIVER FIBROSIS-RENAL TUBULAR ECTASIA SYNDROME

Syndromography

Clínic

This syndrome begins in childhood or young adulthood and is characterized by: portal hypertension manifested by hematemesis and/or melena, hepatosplenomegaly, recurrent attacks of cholangiitis, recurrent attacks of renal colic with urinary infection that can lead to kidney failure.

Paraclinical examinations

Liver biopsy. A dilation of the interlobular bile ducts is observed with a marked increase in fibrous tissue around the portal spaces.

Radiology. Dilation of the renal tubules similar to the so-called spongy renal medulla with or without calcifications. Dilation of the intrahepatic biliary system. Varicose veins esophageal. Signs of intrahepatic portal hypertension.

Syndromogenesis and etiology

This syndrome is hereditary and is probably transmitted in an autosomal recessive manner.

From the etiological point of view, Alagille suggests that it should be considered a dysplasia or alteration at the level of the portal ductal plate.

Literature

Alagille, D. and M. Odrieve: Liver and biliary tract disease in children. New York, Wiley Medical Pub., 1979.

Grossman, H. et al.: "Congenital hepatic fibrosis, bile duct dilation and renal lesion resembling medullary sponge kidney (congenital "cystic disease of the liver and kidneys)." Radiology, 87:46, 1966.

Mayans, J.A., N. Mata, et al.: "Congenital hepatic fibrosis. Study of 26 cases." Acta Gastro Latinoamericana., 24 (5):297- -303, 1994.

Unite, I., et al.: "Congenital hepatic fibrosis associated with renal tubular ectasia: A report of three cases." Radiology. 109:565, 1973.

CYSTIC FIBROSIS SYNDROME - CHRONIC LUNG DISEASE

Synonymy

Cystic fibrosis of the pancreas. S. of multilobar biliary cirrhosis.

Syndromography

Among the most frequent syndromes are: cystic fibrosis-chronic lung disease and multilobar focal biliary cirrhosis (see).

Clinic

Cystic fibrosis-chronic lung disease syndrome is characterized by: abdominal cramps, voluminous and foul-smelling diarrhea, voracious appetite, respiratory symptoms: dyspnea, chronic cold, production of sticky viscous sputum, fever with prostration if the temperature is high.

Physical examination. Malnutrition, delayed growth, delayed puberty, distended abdomen, pulmonary hyperresonance, flat diaphragm, clubbed fingers on occasions.

Paraclinical examinations

Sweat. Increase in electrolytes sodium and chloride and decrease in potassium.

Blood. Normal electrolytes; If exposed to heat, sodium drops with normal potassium levels. During bronchial episodes: increased CO,

Pancreatic secretion. Presence of specific mucoprotein. Deficiency of pancreatic enzymes (trypsin, lipase and amylase).

Stool. Steatorrhea and azotorrhea (nitrogen losses).

Intestinal biopsy. Dilation of the mucous cells of the duodenal villi. Brunner's glands distended and with positive PAS material.

Syndromogenesis and etiology

It is a syndrome of genetic etiology whose gene was located at 7q22.3-q23.1. The mutational defect initially described, known as the delta F 508 mutation, consisted of a deletion of three base pairs, which resulted in the loss of a phenylalanine amino acid residue at position 508 of the gene product. This protein functions as a regulator of transmembrane conductance and its function is affected in CF (CFRT), this causes the clinical picture described here. The clinical heterogeneity of symptoms is due to a large number of other mutations (currently 41 of them have been studied biochemically). The protein is inferred to have 1,488 amino acids and a molecular weight of 168,138 Da, consisting of two sequences, each with domains that have properties consistent with membrane proteins and a domain that is estimated to be related to ATP binding.

Etiological knowledge has allowed the use of gene therapy, in these cases using aerosols with adenovirus to which the normal CFRT protein gene has been transferred. Analysis of lung RNA from patients has revealed the presence of normal gene transcription for a period of up to 6 weeks, notably improving the clinical manifestations of pulmonary cystic fibrosis.

The aforementioned causes a generalized dysfunction of the mucous exocrine glands with the production of abnormally thick mucus and the subsequent obstruction and infection of the bronchi, pancreatic and bile ducts in the form of focal biliary cirrhosis. Ultimately this leads to fibrosis of these structures.

Literature

Di SantAgenese, P.A.: “Cystic fibrosis of pancreas: mucoviscidosis, fibrocystic disease of the pancreas.” Deutsch. Med. Wschr., 86:1376-1384, 1961.

Kerem, B.: J.M. Rommens: J.A. Buchanan, et al.: "Identification of the cystic fibrosis gene: genetic analysis." Science 246:1073- -1080, 1989.

Riordan, JR; J.M. Rommens; B. Kerem, et al.: "Identification of the cystic fibrosis gene: cloning and characterization of complementary DNA." Science, 245:1066-1073, 1989.

Rosenfeld, M.A.; K. Yoshimura; B.C. Traporell, et al.: In vivo transfer of the human cystic fibrosis transmembrane conductance regulatory gene to the airway epithelium." Cell, 68:143-155, 1992.

Zielenski, J.; D. Bozon; B. Kerem, et al.: "Identification of mutations in esfons 1 through 8 of the cystic fibrosis transmembrane conductance regulator (CFTR) gene." Genomics, 10:229-235, 1991.

FITZ SYNDROME

Synonymy

S. of acute hemorrhagic pancreatitis. S. of acute necrosis of the pancreas. S. of acute pancreatitis.

Syndromography

Clinic

Fitz syndrome can occur at any age, but is more common in women. It is characterized by intense pain in the epigastrium with radiation to both sides and especially to the back, which is sometimes not relieved even with opiates. Sometimes it is partially relieved with certain positions: sitting and leaning forward, Cheinise position. There are occasional cramps, nausea, vomiting, constipation, and shock in 45% of cases.

Physical examination. Fever, tachycardia, weak pulse, hypotension, jaundice, cold skin. Abdomen: subcutaneous hemorrhagic phenomena that sometimes occur days before the severe episode and are in the discoloration phase. Grey-Turner sign (hemorrhagic suffusions on the flanks), Cullen sign (periumbilical hemorrhagic sufusion). Pain on palpation of the abdomen with contracture or abdominal guarding (50% of patients). Decrease in intestinal peristalsis that can lead to paralytic ileus in 10% of cases. Chest: presence of signs of pleural effusion or atelectasis, especially at the bases, causing respiratory failure, in 30% of patients.

Paraclinical examinations

Blood Leukocytosis, elevated bilirubin, elevated alkaline phosphatase, hypocalcemia on the second day, which is essential information for the diagnosis. Hyperglycemia at times. Very high serum amylase and lipase in blood and urine.

Paracentesis of pleural or abdominal fluids. They show increased amylase and lipase.

Imaging, ultrasound and computed axial tomography. They can be helpful in demonstrating the increase in gland size.

Laparoscopy. It is the most valuable method for diagnosis. The fundamental element is the cytostatonecrosis lesions, which are round, yellowish-white in color, varying in size between 1 mm and 1 cm, not very prominent and with a reddish halo at their periphery. They are most frequently observed in the greater omentum and the parietal and visceral peritoneum.

Syndromogenesis and etiology

The destruction of the pancreatic acini for various reasons releases the ferments contained therein and causes the damage observed in this syndrome.

It is now accepted that trypsin acts as the primary initiating factor by activating various proenzymes, but is not the direct cause of pancreatic self-digestion.

The activated proenzymes are transformed into the enzymes that we point out below and which are, in turn, the cause of the lesions and symptoms:

1. Elastase: hemolrage.
2. Kalicrein: edema, severe pain and shock.
3. Phospholipase: parenchymal necrosis.
4. Lipase: fat necrosis.

The most frequent causes are: In children, idiopathic or due to infectious diseases. In adults around the fourth decade: due to alcoholism. In the fifth decade: biliary disorders.
Other causes: parasitic infections, hyperparathyroidism, steroid administration, trauma, metabolic, etc.

Literature

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FITZ-HUGH-CURTIS SYNDROME

Synonymy

S. of gonococcal perihepatitis. S. subcostal of Stajano.

Syndromography

Clinic

This syndrome occurs in young women between 20-40 years old. It is characterized by fever at times. Pain in the right upper quadrant, sharp and intense, which raises the diagnosis of an acute abdomen. Often the pain can radiate to the right shoulder and/or back. On other occasions the pain is not as intense but is recurring and resembles pleuritic type pain. Sometimes it is accompanied by nausea without vomiting.

There may or may not be vaginal discharge and pelvic pain or signs of urethritis, and sometimes in some phase of the syndrome the following occur: pharyngitis, salpingitis, pelvic inflammation, etc.

Physical examination. Rub or rub during breathing in the liver region. In more acute cases there is sometimes vaginal discharge and painful vaginal examination.

On physical examination, tenderness is found in the right upper quadrant, adnexa, and cervicitis, even in women whose symptoms do not suggest salpingitis.

Paraclinical examinations

Liver tests. Liver function tests are generally normal.

Ultrasound. Normal right upper quadrant.

Oral cholecystography. Sometimes a deficit in the functioning of the

Laparoscopy. Inflammation of the liver capsule (edema and erythema), presence of exudate with fibrinous adhesions between the visceral and parietal peritoneum. If the examination is performed in late stages due to pain in that area, in untreated cases, "violin string" adhesions may be observed causing persistent pain in the right upper quadrant. These adhesions do not present vascularization and can be sectioned and eliminated during the examination, which definitively resolves the pain.

Vaginal exudate. To identify the bacterial etiology: gonococcus or Chlamydia.

Syndromogenesis and etiology

This syndrome is seen in 5-10% of women with acute pelvic inflammation (API). For years it was attributed to gonococcal IPA, but it is now known that the most important etiological agent is Chlamydia trachomatis.

It is important to note that these adhesions may exist, even if significant adhesions are not seen in the pelvic region or the tubes (origin of the process).

Although it is a primarily gynecological process, upper abdominal symptoms are the attention of the gastroenterologist and the surgeon whom the patients consult.

The mechanism by which the bacteria is capable of producing perihepatitis and adhesions on the upper surface of the liver is discussed.

Literature

Curtis, A.H.: "Cause of adhesion in right upper quadrant." J.A.M.A., 94:1221-1222, 1930.

Fitz-Hugh, T., Jr.: "Acute gonococcal peritonitis of right upper quadrant in women." J.A.M.A., 102:2094-2096. 1934.

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Stajano, C.: The phrenic reaction in gynecology." Sem. Med. B. Aiz., 27:243-248, 1920.

Vickers, EN. and P.J. Maloney: "Gonococcal perihepatitis: Report of three cases with comment on diagnosis and treatment." Arch. Int. Med., 114:120-123, 1964.

COLON HEPATIC FLEXURE SYNDROME

Synonymy

S. of the hepatic flexure.

Syndromography

Clinic

It is characterized by a feeling of abdominal fullness and the desire to burp. Pain in the epigastrium and kidney area (right upper quadrant and lumbar region on that side).

Physical examination. Sometimes tympanism in the right hypochondrium.

Paraclinical examinations

Radiology. It shows gas distension in the affected area and if the x-ray is performed with a barium enema, the bending of the colon at that level is more clearly evident.

Colonoscopy. In many cases it is difficult to exceed the hepatic angle of the colon.

Syndromogenesis and etiology

It is due to the accumulation of gas in the hepatic flexure, of organic or functional cause.

Literature

Palmer, E.D.; D.L. Deutsch and N.M. Scott, Jr.: "Clinical experiences with the splenic flexure syndrome and the hepatic flexure syndrome." Amer. J. Digest. Dis., 22:194-197, 1955.

FRUGONI SYNDROME

Synonymy

S. of splenomegaly-thrombophlebitis of the splenic vein. S. of Cauchois-Frugoni thrombophlebitic splenomegaly. S. of lienophlebitis.

Syndromography

Clinic

Pain in the left hypochondrium, fever and chills. It is accompanied by a portal hypertension syndrome: splenomegaly, hematemesis and melena. The symptoms are similar to those observed in Banti syndrome.

Paraclinical examinations

Radiology or imaging. Checking splenomegaly and/or dilation of the splenic vein.

Laparoscopy. Allows you to specify the site of the obstruction: whether it is primary to the splenic vein, or is secondary to established liver cirrhosis.

Splenography. It is very dangerous.

Syndromogenesis and etiology

It is due to thrombophlebitis with obstruction of the splenic vein that causes splenomegaly. It occurs during infections or surgical interventions near the vein. It can extend to the portal in which case it is accompanied by ascites.

Literature

Frugoni, C.: "La splenomégalie thrombophlebitique." Rev. Belge Sc. Méd., 10:227-236, 1938.

GARDNER SYNDROME

Synonymy

S. of familial multiple polyposis. S. of bone and soft tissue tumors. S. of colic polyposis-epidermoid cysts.

Syndromography

Clinic

This syndrome affects both sexes. It begins at age 20, but polyps can be found from 2 months to 70 years. However, they do not exist at birth. Rectal bleeding is the most common in children. At first it is characterized by mild diarrhea with small amounts of blood and/or mucus, but some patients remain asymptomatic. Between 20-30 years of age, symptoms related to the malignant degeneration of polyps begin.

Physical examination. In soft tissues: multiple epidermoid cysts, desmoid tumors, fibromas, neurofibromas.

In the bones: well-limited benign exostoses, osteomatosis located on the face and skull, impacted supernumerary teeth. Long bones are rarely affected.

Paraclinical examinations

Radiology. Colon by enema to detect polyps, number and location.

Colonoscopy. It is the method of choice and the most accurate, which also allows observing the macroscopic characteristics and/or diagnosing malignant ones and taking a biopsy.

Skin biopsy. In soft tissue tumors.

Fibroblast culture. Increase in tetraploids.

Once the case is diagnosed, the entire family must be examined, since the probability of those with this type of polyps suffering from cancer approaches 100% at the age of 40.

Syndromogenesis and etiology

It is a hereditary disorder, which is transmitted on an autosomal dominant basis, and which has the same gene on which soft tissue and bone lesions and polyps depend.

Polyps are adenomas and their malignancy rate is very high. They frequently also exist in the stomach and duodenum.

The gene has been located on chromosome 5q21, this region contains at least 6 genes that are expressed in the normal mucosa of the colon and encode an unusually large protein.

Literature

Colcock, B.P. and A.A. Zomorodian: "Gardner's syndrome: Multiple polyposis of colon, bone tumors and soft-tissue tumors." Postgrad. Med., 40:29-43, 1966.

Devic, B: “Un cas de polypose adénomatease: généralisée a tout l' intestin'". Arch. Mal. Appa. Dig., 6:278-299, 1912.

Gardner, E.J. and FAITH. Stephens: "Cancer of the lower digestive tract in one family group." Amer. J. Human Genet., 2:41-48, 1950.

GASTROPARESIA SYNDROME

Syndromography

Clinic

This syndrome has been individualized within the motor alterations of the digestive tract and is characterized by chronic nausea, epigastric discomfort and recurrent vomiting with food residues.

Epigastric discomfort is generally defined by patients as a feeling of fullness or pressure, but other times as frank pain. The symptoms are induced

typically (not always) due to the ingestion of especially solid foods. Weight loss and other nutritional disorders are often sequelae that develop over time in severe cases.

Gastroparesis can be classified as a functional disorder and. Therefore, it must be demonstrated, excluding other organic abnormalities of the digestive system such as:
gastric neoplasia, peptic ulcer and inflammatory or parasitic processes of the upper digestive tract.

Paraclinical examinations

Stomach and duodenum radiology. Evidence of delayed gastric emptying.

Radiocintigraphy. It is the most reliable method for verifying the slowdown in gastric evacuation of solid foods.

Manometry. Antral hypomotility. The manometric diagnosis must be performed both on an empty stomach and after ingestion of food.

Ultrasound and impedance techniques. They have a potential advantage because the patient is not exposed to ionizing radiation, but experience with these methods is still limited.

Panendoscopy. It allows us to rule out organic lesions and in experienced hands it can specify the slowness of peristalsis.

Syndromogenesis and etiology

From a pathophysiological point of view, what characterizes the disease is postprandial antral hypomotility, particularly after ingestion of solid foods. It has been established that the following factors participate in the slowing down of gastric evacuation of solid foods: inadequate chewing and deficiency in particle propulsion. Other factors such as: increased pyloric and duodenal resistance caused by uncoordinated contractions in these segments, may also play a role in the pathophysiology of this syndrome.

Gastroparesis can be of primary origin (idiopathic), or be secondary to specific disorders such as: diabetic visceral neuropathy, gastric surgery (vagotomy with or without partial gastric resection) and less common causes such as dysautonomous disorders, collagen and smooth muscle diseases, and paraneoplastic syndromes.

Literature

Malagelada, J.R.: Motility disturbances underlying gastroparesis." In Heading, R.C., J.D. Wood, ed. Gastrointestinal dysmotility.
Focus on Cisapride. Raven Health Care Communications. New York, 1992.

GILBERT SYNDROME N.A.

Synonymy

S. of benign unconjugated bilirubinemia. S. of chronic low-intensity hyperbilirubinemia. S. of juvenile intermittent jaundice. S. of hereditary non-hemolytic bilirubinemia. S.
of familial non-hemolytic jaundice. S. of constitutional liver dysfunction. St. of Meulengracht. S. de Gilbert-Lereboullet.

Syndromography

Clínic

Since Gilbert's original description in 1907, this benign, chronic, characterized by mild, permanent unconjugated hyperbilirubinemia persistent

It affects both sexes. Male-female ratio 4:1. It begins early after birth, but is generally not recognized for several years. There is slight scleral jaundice which is the only finding.

During its evolution, the following may appear: asthenia, nausea, pain in the right upper quadrant. Jaundice increases after alcohol ingestion, intercurrent infections, exertion and prolonged fasting.

Paraclinical examinations

Blood. Unconjugated hyperbilirubinemia. Normal transaminases and alkaline phosphatase. No evidence of hemolysis or other condition that could explain the hyperbilirubinemia. Delayed clearance of bilirubin in the blood. Rapid response to phenobarbital with fall in bilirubin.

Urine. Does not contain bilirubin.

Cholangiography. Normal.

Liver biopsy. Normal.

Syndromogenesis and etiology

Its transmission by autosomal dominant inheritance is considered possible, with incomplete penetrance and variable expressivity.
There appears to be a defect in the uptake of unconjugated bilirubin from serum by the hepatocyte membrane. It is currently proposed that there is a certain deficiency of glucuronyl transferase, and that in some cases a certain degree of hemolysis coexists. For this reason, some authors describe two varieties of Gilbert syndrome: with or without hemolysis.

Two types have also been related: one with normal distribution of the neutral o-glucosidase in which the smooth endoplasmic reticulum is normal and another with abnormal distribution of the indicated enzyme and hypertrophy of the smooth endoplasmic reticulum.

The etiology is unknown

Literature

Gilbert, N.A. and P. Lereboullet,: "La cholemie simple familiale."
Sem. Med., 11:241-243. 1901.
Powell, L.W.; E. Hemingway: B.H. Billing and S. Sherlock:
"Idiopathic unconjugated hyperbilirubinemia (Gilbert's syndrome)". N. Engl. J. Med., 277:1 108-1112, 1967.

HANOT SYNDROME

Synonymy

S. of primary intrahepatic cirrhosis. S. of cholangitic biliary cirrhosis. Chronic destructive non-suppurative cholangitis.

Syndromography

It is necessary to make some prior considerations in this syndrome. Until 1875, the known cirrhosis was the classic Laennec cirrhosis, with atrophic liver, splenomegaly, ascites, collateral circulation and final liver failure with coma. At that time, Hanot studied in the clinic and verified in cadavers another type of cirrhosis, which was characterized by hepatomegaly during almost its entire evolution, sustained jaundice, pruritus and without ascites or manifest splenomegaly.
This condition was known as Hanot's cirrhosis. In all these years, biliary cirrhosis was considered synonymous with Hanot syndrome. In the 20th century, knowledge of this entity advanced and two different conditions were separated: a, cirrhosis

primary biliary, true Hanot syndrome, since he did not find any cause in the cases he describes; b, secondary biliary cirrhosis when there is chronic obstruction and/or infection of the main bile ducts in a chronic form, or stenosis (sclerosing cholangiitis, carcinoma of the walls of the main bile duct, chronic pancreatitis) and in children bile duct atresia.

We will refer to chronic destructive non-suppurative cholangiitis, Hanot syndrome.

Clínica

This syndrome predominates in young or middle-aged women.

age.

It begins insidiously with pruritus, steatorrhea, good general condition that deteriorates and progresses towards a state of malnutrition with malabsorption of fat-soluble vitamins (A, D, K). Mane and in the final stage coma.

Physical examination. Sustained jaundice with greenish discoloration, skin pigmentation, xanthelasma, flat or tuberous xanthomas, especially on the palms and elbows. Generalized lymphadenopathy, hepatomegaly and moderate splenomegaly. Ascites and liver failure in the final stages accompanied by atrophic liver.

Paraclinical examinations

Blood. Increase in bilirubin, lipids and globulins.

Circulating antimitochondrial antibodies of the IgG type in more than 95% of patients. Furthermore, increases in IgM and cryoproteins consisting of immune complexes capable of activating the alternative complement pathway by 80-90% are discovered.

Urine. Darkened by bilirubin and urobilin.

Stool. Discolored with increased fatty acids.

Cholangiography. Absence of extrahepatic obstruction.

Bone radiology. Osteoporosis and late osteomalacia.

Laparoscopy. There is hepatomegaly of a more or less intense green color and a fine granular surface. There are no signs of portal hypertension and the spleen is slightly enlarged.

As it progresses, cirrhosis forms with more or less atrophy, composed of small or medium-sized nodules, although the liver remains green, the opposite of other types of cirrhosis. Already at this stage, signs of portal hypertension appear.

Liver biopsy. The findings depend on the evolutionary stage of the process and four evolutionary stages are recognized.

Syndromogenesis and etiology

It tends to be considered among diseases due to immunological alterations. It frequently accompanies various conditions of a supposedly autoimmune nature, such as CREST syndrome (calcinosis, Raynaud, sclerodactyly, telangiectasia), sicca syndrome (dryness of the ocular and oral mucosa), autoimmune thyroiditis and renal tubular acidosis.

The discovery of the antimitochondrial antibody in its two varieties is another element in favor of this possible pathogenesis.

Four mitochondrial autoantibodies are currently known.

The etiology is unknown.

Literature

Hanot, V.: "Etude sur une forme de cirrhose hypertrophique du foie (cirrhose hypertrophique vecctere chronique)". Medical Classics, Paris, 1875 (Thesis).

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Sherlock, S.: "Primary biliary cirrhosis (chronic intrahepatic obstructive jaundice) ". Gastroenterology, 37:574-586, 1959.

HELLP SYNDROME

Synonymy

S. of hemolysis-elevated liver enzymes-low platelet count. (H: hemolysis; EL: enzymes liver; LP: low platelet).

Syndromography

Clínic

This syndrome occurs in the second half of pregnancy and suggests severe liver disease in a patient with hypertension during pregnancy.

High blood pressure has always been present for months, although patients may be asymptomatic;
However, many of them present pain in the right upper quadrant or epigastrium accompanied by the clinical manifestations of a mild hemolytic syndrome.

Paraclinical examinations

Blood. Elevation of aminotransferase levels with values that can range between 60 and 1,500 units.

Platelet count: decreased, less than 100,000.

Liver biopsy. Periportal hemorrhage and fibrin deposits. Sometimes fatty infiltration.

Periportal hemorrhage and fibrin deposits do not correspond to the degree of steatosis. The liver does not show the fatty microvesicular arrangement typical of acute steatosis of pregnancy, suggesting that these are two different entities from the point of view of their histology.

There is also no correlation between the severity of the histological damage, the level of liver enzymes in the blood and the patient's clinical manifestations.

Syndromogenesis and etiology

The exact pathophysiological mechanism of this syndrome is not known, but the presence of high blood pressure in all cases suggests that it is related to it in some way, perhaps due to the hypoxia that causes or may cause this alteration in the liver cells.

The etiology is unknown.

Literature

Barton, JR; AC Riely: T.A. Adamec, et al,: "Hepatic histopathologic conditions does not correlate with laboratory abnormalities in HELLP syndrome (hemolysis, elevated liver enzymes and low platelet count)." Am. J. Obstet. Gynecol., 167:1538-1543. 1992.

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Weinstein, L.: “Syndrome of hemolysis, elevated liver enzymes and low platelet count: a severe consequence of hypertension in pregnancy". Am. J. Obstet. Gynecol., 142:159-167. 1982.

RADIATION HEPATITIS SYNDROME

Syndromography

Clínic

It appears approximately 4 weeks after radiotherapy to the liver area. The clinical picture is reminiscent of

Budd-Chiari syndrome (see) and is characterized by jaundice, ascites, pain in the right upper quadrant and rapid onset hepatomegaly.

Paraclinical examinations

Blood. Alkaline phosphatase is increased.

Ultrasound, computerized axial tomography and selective arteriography. They should be performed to check the integrity of the hepatic veins.

Liver biopsy. Minimal changes are seen in the liver parenchyma. Damage to the venules can cause congestion and stasis.

Syndromogenesis and etiology

This syndrome is due to the harmful action of radiation on the structures of the liver.

Literature

Ingold, J.A.; G.B. Reed and H.S. Kaplan, et al.: "Radiation hepatitis." Amer. J. Roentg., 93:200-208, 1965.
Lansing, A.M.; W.M. Davis and H.E. Brizel: "Radiation hepatitis." Arch. Surg., 96:878-882, 1968.

HEPATOCEREBRAL SYNDROME

Synonymy

S. of postshunt encephalopathy (post-derivation). S. of portosystemic encephalopathy. S. of Gaustad. S. of transient hepatargia. Neuropsychiatric S. of liver cirrhosis.

Syndromography

Clínic

The clinical picture is characterized by: mental confusion, apathy, dizziness, delirium, alteration of behavior and character, personality change, hypersomnia, dysarthria, headache, blurred vision.

Physical examination. Precoma state without loss of consciousness, hypertonia with hyperreflexia, flapping tremor in the hands, fingers and wrist. Asterixis is very common. There is palmar erythema, spider veins, loss of body hair.
Additionally, signs of portal hypertension (collateral circulation, splenomegaly).

When flaccid coma is established, areflexia in the limbs, jaundice, breath or hepaic fetor are observed.

Paraclinical examinations

Blood. Mild hyperbilirubinemia, prolonged prothrombin time, hypoproteinemia with hypoalbuminemia, increased blood nitrogen, increased alkaline phosphatase, hypocalcemia, hypokalemia, hyponatremia, hypoglycemia.

Electroencephalogram. There are alterations in the frequency and amplitude of the waves.

Syndromogenesis and etiology

Symptoms of liver failure occur along with disorders due to elevated blood nitrogen. The precipitating causes of this condition are: anoxia, intercurrent infections, pregnancy, abdominal paracentesis, increased intestinal ammonia due to hemorrhage and/or bacteria, blood transfusions, drugs, surgery, hypokalemia, shunt, alkalosis, increased protein in the diet. Hypokalemic alkalosis is due to diuretics.

The most important factors are: severe hepatocellular dysfunction and extrahepatic or intrahepatic shunting of portal blood into the systemic circulation bypassing the liver, or both. Consequently, toxic substances absorbed from the intestine are not detoxified in the liver and cause metabolic alterations in the central nervous system.

Although ammonia is considered the most important cause of brain alterations, other substances have been considered as propitiators of this phenomenon, they are: mercaptans (derived from the metabolism of methionine), short chain fatty acids, phenol and gamma-aminobutyric acid (GABA), which is an inhibitory neurotransmitter. False chemical neurotransmitters such as octopamine, a product of the metabolism of aromatic and branched chain amino acids, may also participate.

Neuropsychic manifestations are reversible when the encephalopathy improves. This does not happen in terminal cases with liver failure.

Literature

Baker, A.B.: "Interrelationship of diseases of the liver and brain." Arch. Path., 46:268-286, 1948.
Hahn, M.: 0. Massen; M. Wencki, et al.: "Die Eck'sche Fistel Zwischen der Unteren Holivene und der Pfortader und ihre Folgen fur den Organisms." Arch. Exp. Path. Lpz., 32:151, 1893.

McDermot, W.V.: B.A. Barnes: G.L. Nardi, et al.: “Postshunt encephalopathy.” Surg. Gynecol. Obstet., 126:585-590, 1968.

HEPATORRENAL SYNDROME (HRS)

Synonymy

S. de Heyd. S. urohepatic. Hepatourological S. S. of cholemic nephrosis.

Syndromography

Clinic

All kidney syndromes that occur in patients with liver conditions are considered here. The clinical picture combines the symptoms of both conditions: jaundice, oliguria-anuria, irritability, etc.

It is a serious clinical condition, since the symptoms of liver injury are associated with kidney symptoms with retention of urea, creatinine, sodium, etc., related to marked oliguria.

From a clinical point of view, there are two types of HRS that are differentiated by their different clinical evolution and prognosis.
HRS type I is characterized by rapidly progressive renal failure defined by a 100% increase in the initial serum creatinine level to a level greater than 2.5 mg/dL (221 umol/L) or a 50% reduction in the initial 24-h creatinine clearance to a value less than 20 mL/min in a period of less than 2 weeks. In HRS type II, renal failure is stable or does not meet the progression criteria defined for type I.

Patients with HRS type I have a median survival of less than 2 weeks from diagnosis, while patients with HRS type II have longer survival.

Paraclinical examinations

Liver and kidney function tests. They are altered.

The determination of pressure in the inferior cava is closely correlated with glomerular filtration pressure and urinary sodium excretion.

Diagnostic criteria:

Unfortunately, there is no clinical or analytical parameter that allows the diagnosis of HRS to be established with certainty.

For this reason, the diagnosis must be established according to a set of criteria whose fundamental objective is to exclude other causes of renal failure that can frequently be observed in patients with chronic liver disease, especially those secondary to shock, dehydration, bacterial infections, administration of nephrotoxic drugs (non-steroidal anti-inflammatory drugs, aminoglycosides, etc.) or glomerulonephritis due to immune complex deposits.

The diagnostic criteria for hepatorenal syndrome according to the International Ascites Club*;

Major criteria are:

1. Acute or chronic liver disease with advanced liver failure and portal hypertension.
2. Low glomerular filtration rate, estimated by serum creatinine > 1.5 mg/ dL (133 umol/L) or 24-h creatinine clearance <40 mL/min.
3. Absence of shock, active bacterial infection and current or recent treatment with nephrotoxic drugs. Absence of fluid losses through the gastrointestinal tract (repeated vomiting or severe diarrhea) or kidney (weight loss > 500 g/day for several days in patients with ascites without peripheral edema > 1,000 g/day in patients with peripheral edema).
4. No improvement in renal function (decrease in serum creatinine to a value of 1.5 mg/dL (133 umol/L) or increase in creatinine clearance to a value of 40 mL/min) after withdrawal of diuretic treatment and expansion of plasma volume with 1.5 L of isotonic saline solution.
5. Proteinuria < 500 mg/day.
6. Absence of ultrasound signs suggestive of chronic kidney disease or obstructive uropathy.

Minor criteria:

1. Very low urine output.
2. Urinary sodium < 10 mEq/L.
3. Urinary osmolarity > plasma osmolarity.
4. Red blood cells < 50/field.
5. Serum sodium concentration < 130 mEq/L.

Syndromogenesis and etiology

Hepatorenal syndrome is a syndrome that occurs in patients with chronic liver disease, generally liver cirrhosis or alcoholic hepatitis, with advanced liver failure and portal hypertension, which is characterized by a marked alteration of renal function and anomalies in arterial circulation and in the activity of endogenous vasoactive systems.

At the renal level there is an intense arterial vasoconstriction that determines a very significant reduction in renal blood flow and glomerular filtration, in the absence of histological abnormalities.

There is also intense renal retention of sodium and water, responsible for the formation of ascites, edema and, frequently, dilutional hyponatremia.

Unlike what occurs in the renal circulation, arterial vasodilation predominates in the extrarenal circulation, which causes a decrease in systemic vascular resistance and arterial hypotension.

Hepatorenal syndrome can also be observed in patients with liver diseases that present with acute liver failure.

The mechanism of kidney failure is unknown. However, every effort should be made to rule out other conditions that are accompanied by liver and kidney dysfunction, such as:

1. Infections (leptospirosis, immune complex hepatitis).
2. Toxic (aminoglycosides, carbon tetrachloride).
3. Circulatory disorders (shock, severe heart failure).

It is important to remember that jaundiced patients with liver disease may be particularly susceptible to developing acute tubular necrosis, which should not be confused with this syndrome.

There are no renal histological alterations in these patients and when kidneys from these patients are transplanted to individuals without liver failure, the function is normalized, which suggests the presence of a functional defect. On the other hand, the improvement in liver function is accompanied by improvement in kidney function.

Literature

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REDUCIBLE HERNIA SYNDROME

Syndromography

Clínic

This syndrome is important from a clinical point of view, as it can simulate various abdominal symptoms that lead to false interpretations. These paintings are:

*All major criteria are necessary for the diagnosis of hepatorenal syndrome. The minor criteria are not essential, since their existence reinforces the diagnosis

1. Symptoms that simulate peptic ulcer.
2. Symptoms that simulate irritable colon.
3. Symptoms that simulate gastritis.

Syndromogenesis and etiology

The etiology is a reducible hernia that causes reflex manifestations that are sometimes distant from the site of origin. The reflex mechanism is caused by transient compression of the intestine and/or hernial sac. When a hernia becomes stuck and/or strangulated, the clinical picture is clearer and lends itself to less confusion.

HYPERSPLENISM SYNDROME

Synonymy

S. hypersplenic. S. splenic.

Syndromography

Clinic

It is basically characterized by: anemia, leukopenia and thrombocytopenia in the peripheral blood that contrasts with a normal or, generally, hypercellular medulogram. It is almost always accompanied by splenomegaly. However, it is not necessary for there to be pancytopenia, since there can basically be a decrease in only one of the cellular elements.

The criteria for the diagnosis of hypersplenism include:

1. Splenomegaly.
2. Splenic destruction of one or more cell lines in peripheral blood.
3. Normal or hyperplastic cellularity in the bone marrow with normal representation of the deficient cell line in the circulation.
4. Signs of increased turnover in affected cell lines, for example, reticulocytosis, increased banded neutrophils, or circulating immature platelets.

Syndromogenesis and etiology

In hypersplenism the spleen eliminates excessive amounts of erythrocytes, granulocytes or platelets from circulation. The general mechanisms of elimination of formed elements from the blood fundamentally comprise greater cellular sequestration, due to hemodynamic abnormalities in the splenic circulation or due to the production of antibodies against erythrocytes, granulocytes or platelets that make these cells more vulnerable to phagocytosis by splenic macrophages.

States in which there is passive congestion of the spleen produce abnormal sedimentation of blood in sinuses and cords of the red pulp. Under these conditions there is a storage of plasma that produces notable intrasplenic hemoconcentration and hypoxia, which makes the blood cells more vulnerable to the phagocytic action of macrophages from the pulp cords.

PORTAL HYPERTENSION SYNDROME

Syndromography

Clinic

The main clinical manifestations of portal hypertension are: esophagogastric varices, ascites, collateral circulation, splenomegaly with or without hypersplenism, symptoms of acute or chronic hepatic encephalopathy.

All of these symptoms are related, in part, at least to the formation of a portosystemic collateral circulation.
The absence of valves in the portal system facilitates retrograde circulation (hepatofugal) from the hypertensive portal system to where there is less pressure. The main sites of collateral circulation are: in the veins of the rectum (hemorrhoidal), cardioesophageal junction (gastroesophageal vaices), retroperitoneal space and falciform ligament of the liver (periumbilical and/or abdominal wall veins). Sometimes these wall vessels are observed in the epigastrium as tortuous vessels that are arranged in a radiating pattern starting from the navel towards the xiphoid appendix and costal edge (head of the medusa).

Paraclinical examinations

Radiology. Barium esophagram confirms varicose veins esophageal.

Splenoportography. To detect the site of intrahepatic or extrahepatic obstruction.

Portal pressure measurement. Directly with a fine needle or indirectly by inserting a cannula into the suprahepatic veins. Whenever you decide to do this test, the free and wedge pressure should be measured to determine if it is a presinusoidal (normal pressure), sinusoidal and post-sinusoidal (elevated pressure) obstacle.

Esophagoscopy. Specifies the existence of varicose veins and their intensity and extent.

Laparoscopy. It allows you to check whether it is an injury above the liver (Bud-Chiari syndrome), injury to the liver (liver cirrhosis), or injury below the liver (cavernomatosis or portal angiomatosis, compressions, etc.). In addition, it reports on internal collaterals in sites that are not accessible to physical examination.

Syndromogenesis and etiology

Portal pressure is usually low (10-15 cm saline; 7-10 mm Hg) because vascular resistance in the hepatic sinusoids is minimal. Portal hypertension (>30 cm of saline) depends on increased resistance to portal flow. Since the portal system does not have valves, resistance at any level between the heart and the splanchnic vessels produces hypertension by retrograde transmission. This increased resistance can occur at three levels in relation to the hepatic sinusoids:

1. Presinusoidal.
2. Sinusoidal.
3. Postsinusoidal.

Obstruction in the presinusoidal venous compartment may be extrahepatic (e.g., portal thrombosis, cavernomatosis) or hepatic, but functionally proximal to the sinusoids, so that the parenchyma is not exposed to venous hypertension (e.g., in schistosomiasis).

Sinusoidal lesions are the most common and are mainly due to liver lesions, especially liver cirrhosis.

Postsinusoidal occlusion can occur outside the liver at the level of the suprahepatic veins (for example, in Bud-Chiari syndrome), inferior cava. Less commonly, it may be hepatic (e.g., venocclusion of the centrilobular venules as the primary site of injury).

The distinction between these three levels is often interesting, but functional resistance in a given patient may also occur at more than one level.

Portal hypertension can also be caused by increased blood flow (for example, massive splenomegaly).

or arteriovenous fistulas), but it is a rare problem in the clinic due to the little resistance that the blood encounters when leaving a normal liver.

In practice, the most common cause of portal hypertension is liver cirrhosis.

HIRSCHPRUNG SYNDROME

Synonymy

S. of congenital megacolon. S. of aganglionic megacolon.

Syndromography

Clínica

Prevalence in men 3:1. It starts from birth. The intensity of symptoms depends on the extent of the aganglionic segment. It is characterized by: obstipation, vomiting, rapid dehydration. Small amounts of flattened stools may eventually be passed. In some cases sporadic diarrhea may be the main symptom. In untreated cases there is developmental deficit and weakness.

Physical examination. Depending on the size and location of the affected colonic segment there may be: increased peristalsis, abdominal distension, elevation of the diaphragm. Masses of feces (fecalomas) are palpable in the lower abdomen and are mobile and depressible. Rectal examination shows normal anus and rectum, the latter generally empty; sometimes write them down. Fecal impacts can be palpated in the anterior part of the rectal blister.

The most frequent complications are: fecal impact, perforation, ulceration and hemorrhage.

Paraclinical examinations

Sigmoidoscopy. Normal distal segment. Other parts dilated and filled with impacted feces. Reddish mucosa with small ulcerations.

Rectal biopsy. Shows aganglionosis in the affected area.

Radiology. Narrowing of an area of the colon with marked proximal dilation.

Syndromogenesis and etiology

There is no defined hereditary pattern although familial cases have been reported.

The symptoms are due to a well-defined narrowing of a segment of the sigmoid colon, which causes hyperperistalsis with muscular hypertrophy and proximal dilation of the normal colon, this can produce fecal stasis, impaction, etc.

Polygenic or multifactorial inheritance is proposed; for cases with aganglionosis in the area beyond the sigmoid colon, inheritance compatible with autosomal dominant; For cases that do not go beyond this area, multifactorial or autosomal recessive inheritance with low penetrance.

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LIVER FAILURE SYNDROME

Liver failure can be defined as a failure in liver functions. When it occurs during the course of acute liver disease, it is called acute liver failure and when it occurs in the advanced stages of chronic liver disease, it is considered chronic liver failure.

Both liver diseases have many common elements, both clinically and functionally, and in recent years there have been advances and new concepts that justify expanding these topics, although we need to expand more than usual in the style of this work.

Syndromography

Clínica

The diagnosis of this syndrome is eminently clinical and the concept of liver failure is functional. The causes of this syndrome are very varied, so there is no characteristic histopathological image.

Chronic liver failure comprises a group of clinical manifestations, such as the general condition that can be more or less marked depending on the evolutionary stage of the underlying disease, and which is manifested by asthenia, easy fatigue, anorexia, weight loss in relation to the loss of muscle mass, which is more evident in the limbs and in the temporomaseterine region, as well as in the thenar and hypothenar eminences of the hands. Jaundice may not occur or may be mild, as it depends on the balance between necrosis and liver regeneration; Its presence is an index of disease activity and constitutes an element of poor prognosis.

The skin shows the presence of spider veins or arachnoid telangiectasias, most frequently located in the upper regions of the chest, shoulders and behind the ears. There is palmar erythema in the tenare hypothenar regions and the fingertips at the level of the last phalanx. In men, body hair is reduced or disappears and the pubic hair resembles the female pattern. There may be other manifestations of feminization such as: gynecomastia and testicular atrophy.

In women, oligomenorrhea, loss of breast and pelvic fat, and infertility occur.

Neuropsychic manifestations are common and characteristic, constituting the picture of hepatic encephalopathy or portosystemic encephalopathy, which begins with slight mental alterations, which can only be evidenced by tests of intellectual capacity. Later, more obvious symptoms appear, ranging from irritability to frank psychosis.

Emotional disorders are characteristic: euphoria, depression, apathy, confusion, behavioral disorders, inversion of the sleep pattern, disorientation, lethargy, bradylalia and finally coma, which can reach different degrees of depth or be reversible.

A typical neurological sign, but not pathognomonic of this syndrome, is asterixis, fluttering or trembling of the limbs in certain positions. Extrapyramidal signs can also be detected, such as: rigidity due to increased muscle tone and the cogwheel sign, as well as hyperreflexia and bilateral Babinski sign.

This condition of portosystemic encephalopathy occurs recurrently in stable cirrhotics, either spontaneously without apparent cause or secondary to destabilizing factors, more frequent in patients undergoing surgery.

portacaval shunt. On other occasions this picture of encephalopathy manifests itself chronically, permanently, following an oscillating course in intensity, but progressive and irreversible.

Of great value is the detection of hepatic breath or fetor hepatico, which has been described as a sweet or fruity odor in the breath of many patients and which is related to the severity of liver failure.

In acute and chronic recurrent hepatic encephalopathy, it is relatively easy to recognize four degrees of intensity according to the progressive presentation of neuropsychic signs:

Stage I. Loss of affectivity, euphoria, depression, apathy, anxiety, mild confusion, lack of perception, shortened attention span, motley behavior, altered sleep pattern, asterixis, slurred speech, constructive apraxia, difficulty writing.

Stage II. Lethargy, drowsiness, disorientation, moderate confusion, hepatic fetor, asterixis.

Stage III. Intense confusion, incoherent speech, deep drowsiness (but can wake up), response to stimuli, hepatic fetor, asterixis, grasping and sucking reflex, limb rigidity, hyperreflexia and clonus.

Stage IV. Deep coma (does not respond to stimuli), hepatic fetor, decreased reflexes, muscle hypotonia.

The presence of hemorrhagic diathesis in liver diseases is rare if one takes into account that 75% of severe liver diseases have an abnormal coagulation mechanism and 50% of patients with cirrhosis have increased fibrinolytic activity; However, hypocoagulability can cause severe bleeding after trauma or diagnostic procedures such as liver biopsy. Spontaneous hemorrhagic diathesis only occurs when prothrombin group factors and factor V are below 20% of their normal value, which occurs in severe hepatitis and in the final stages of liver cirrhosis. Hemorrhagic diathesis is also observed when other disorders are associated such as:

intense thrombocytopenia, evident fibrinolytic activity or disseminated intravascular

Acute liver failure appears as a consequence of acute massive or submassive necrosis of the liver, or due to a sudden and severe deterioration in the functions of liver cells.

A study was recently conducted at Kings College Hospital in London and several concepts about acute liver failure syndrome were defined. The term hyperacute liver failure was proposed for cases in which encephalopathy occurs within 7 days after the onset of jaundice. The term acute liver failure was suggested for cases in which the interval between jaundice and encephalopathy ranges between 8-28 days; and the concept of subacute liver failure should be applied when encephalopathy occurs between 5-12 weeks from the onset of jaundice.

Other authors use the terms fulminant and subfulminant liver failure, the first in those patients who develop encephalopathy within 2 weeks following the onset of jaundice and the second to describe cases in which the interval is between 2-12 weeks.

For most hepatologists, the concept of acute liver failure implies the presence of hepatic encephalopathy. However, others extend the concept to all cases of acute liver disease in which the concentration of prothrombin or factor V is less than 50% of normal and use the terms fulminate or subfulminate as soon as the encephalopathy is established.

The absence of a history of previous liver disease has always been considered a requirement for presenting with acute liver failure; however, it is currently known that acute liver failure is one of the ways in which Wilson's disease and chronic active autoimmune hepatitis present.

The diagnosis of acute liver failure syndrome is based on the presence of the two main manifestations of acute liver failure: hepatic encephalopathy and a decrease in the prothrombin rate below 50%.

The clinical picture usually begins suddenly, due to: jaundice, establishment of hepatic encephalopathy, followed by the rest of the symptoms and signs described in the chronic form and the biological alterations that we will describe later.

It is worth highlighting the extreme rapidity with which encephalopathy can go from light degrees to deep degrees, which can lead to coma in a few hours.

A very important finding in the physical examination is the reduction in the size of the liver, which sometimes implies the disappearance of hepatic dullness.

Acute liver failure causes the appearance of the following complications, which are, of course, part of the clinical picture:

1. Bleeding, usually digestive, spontaneously or at the slightest trauma or diagnostic procedure.
2. Renal failure, in more than half of patients, either functional renal failure or acute tubular necrosis.
3. Bacterial infections, generally caused by gram-negative bacilli and that affect the respiratory tract, kidneys or are generalized.
4. Respiratory failure due to infections or respiratory distress.
5. Cerebral edema, especially in young patients with grade IV encephalopathy, which is manifested by tonic-clonic seizures, generalized muscle rigidity, high blood pressure, significant hyperventilation and hyperthermia without apparent cause. The fundus is, in general, normal.

Paraclinical examinations

Blood. Aminotransferases and bilirubin. There is a very peculiar behavior among them, which is due to the anatomical lesion (extensive necrosis of the hepatocytes), as occurs in fulminant hepatitis with massive or submassive necrosis. In these cases, the aminotransferases are very high at the beginning of the clinical picture, with frequent inversion of the ASAT/ALAT index, but over time the values decrease rapidly and this drop is accompanied by a notable increase in bilirubin at the expense of the direct or conjugated fraction.

Coagulation factors. Decrease in plasma concentration, as an expression of failure in the synthesis function, especially of factors I, I, V, VII, IX and X. This leads to a prolongation of the prothrombin time.

Plasma proteins. Hypoalbuminemia, associated with hyperglobulinemia, with inversion of the serine/globulin ratio (characteristic finding of chronic liver failure); or hypoalbuminemia alone, typical of acute liver failure.

Hypoglycemia. Important alteration in these cases.

Ionogram and gasometry. Electrolyte and acid-base balance disorders. The most important are: hypokalemia and respiratory alkalosis, although in the most severe cases metabolic acidosis appears.

Ammonia. Elevated in blood, although there is no correlation between the degree of elevation and the intensity of the encephalopathy.

Electroencephalogram. Progressive disappearance of normal alpha activity with the appearance of slow activity with triphasic waves.

Syndromogenesis and etiology

Liver failure is due to the loss of almost all liver functions, which results in major biological alterations, since the liver has a large number of functions that affect the metabolism and synthesis of numerous substances, essential for life.

Alterations in carbohydrate metabolism are the cause of hypoglycemia, the origin of which lies in the scarce glycogen reserves that the liver can store under these conditions, the poor capacity for neoglycogenesis, and perhaps a deficit in the degradation of circulating insulin.

The synthesis function is notably compromised, which causes a deficiency in coagulation factors, especially fibrinogen and prothrombin. Albumin is the main plasma protein synthesized by the liver; Its concentration is decreased, but since its half-life is relatively long, it will be markedly decreased in chronic liver failure and not in acute liver failure.

Portosystemic encephalopathy is due to hepatic or extrahepatic shunts, through which portal blood passes into the systemic circulation without being cleared by liver cells. In patients with acute liver failure, the short circuit occurs through the liver itself due to the large number of destroyed hepatocytes, incapable of metabolizing the contents of the portal blood. In this way, the blood passes without being modified to the suprahepatic veins and from there to the general circulation.

Therefore, in these patients, liver failure is the main cause of encephalopathy.

In patients with liver failure in chronic liver diseases, such as cirrhosis, portal hypertension causes the formation of collateral veins that connect the portal circulation with the systemic circulation without passing through the liver, in a large proportion. In this case, portal hypertension is the main pathophysiological mechanism of encephalopathy.

The passage of blood from the portal vein to the systemic circulation through the short circuits described above, without passing through the liver cells, prevents the detoxification of many exogenous or endogenous substances absorbed in the intestine that are toxic to the body and especially to the brain, and this is the pathophysiological basis of hepatic encephalopathy.

A large number of potentially harmful substances have been described, however, the real leading role of each of them remains highly debated. In fact, in many of them it is not clear whether these substances constitute the cause or are the consequence of encephalopathy. The chemical substances or products most considered in this aspect are the following:

Ammonium. It has been pointed out as one of the most important factors in the pathogenesis, which is why many authors call this condition hepatoammoniacal encephalopathy. There is clinical and biological evidence for this assertion; however, hyperammonemia does not explain all the causes of hepatic encephalopathy.

Mercaptans. Methanethiol, etanethiol and dimethyl sulfoxide are products derived from methionine by the action of colonic bacteria. They are metabolized by the liver and in cases of cirrhosis, they can reach high plasma levels.
Methionine is known to cause hepatic encephalopathy both clinically and experimentally, and hepatic fetor is produced by a mercaptan derivative. However, there is experimental evidence against the pathogenic effect of these substances.

fatty acids. An increase in the plasma concentration of short or medium chain fatty acids has been proven in patients with liver failure and in Reye's syndrome.

Very high concentrations of these substances have been found, at levels that cause coma in experimental animals. However, it has not been possible to correlate the degree of encephalopathy with the plasma levels of these products.

Amino acids and theory of false neurotransmitters. In patients with liver cirrhosis, there is a characteristic pattern of plasma amino acids: under normal conditions the molar ratio of branched chain amino acids (BCA) (valine, leucine, isoleucine) and aromatic amino acids (AA) (tyrosine, phenylalanine and tryptophan) is 3 to 2 and is reversed in cirrhotic patients. Phenylalanine is converted to phenylethanolamine and tyrosine to ectopamine. These substances cross the blood-brain barrier, reach the central nervous system and act as false nervous transmitters, there they displace the true neurotransmitters, thus causing alterations in the conduction of the nervous stimulus, since the power of these false neurotransmitters is up to 500 times less than dopamine and norepinephrine; It is proposed that this is the neurobiochemical substrate that causes the clinical manifestations of hepatic encephalopathy. This hypothesis, put forward years ago and which is logical, has had contradictions in light of subsequent studies in experimental animals and in therapeutic trials.

GABA (gamma aminobutyric acid). GABA is an inhibitor of synaptic transmission produced by bacteria in the colon. Experimental evidence supports the role of this substance in encephalopathy and an increase in neuronal receptors for GABA has been found. These receptors are molecular complexes that contain binding sites for benzodiazepines and barbiturates, which interdependently can produce a neuroinhibitory effect. The increase in the number of these receptors could explain the hypersensitivity to these drugs.

The fact that it has been demonstrated that GABA does not cross the blood-brain barrier casts doubt on the pathogenic role of this substance in hepatic encephalopathy.

Endogenous benzodiazepines. It is being considered that there are benzodiazepines of endogenous origin that would have an agonist effect on the receptors for GABA and benzodiazepines, giving rise to a neuroinhibitory effect.

Zinc. Alteration of zinc metabolism is of particular importance in the pathogenesis of encephalopathy. This metal
It is involved in two metabolic pathways for the elimination of ammonium: it is necessary for the enzyme ornithinetranscarbamylase of the ureogenesis cycle and for glutaminosynthetase (an enzyme that incorporates ammonium into glutamate).

The most recent opinions seem to favor the view that it is not a single substance or a single altered metabolic process that explains the pathogenesis of hepatic encephalopathy.
It has been shown that there is a synergism between ammonium, mercaptans and short chain fatty acids associated with electrolyte disorders, hypoxemia, hypoglycemia and bacterial toxemia.

In conclusion, it can be stated that hepatic encephalopathy is currently considered to have a multifactorial pathogenesis.

Regarding etiology, liver failure can occur in practically any type of acute or chronic liver disease. In the case of chronic liver diseases, liver cirrhosis and chronic active hepatitis are the two entities that cause the vast majority of these syndromes.

There are numerous causes of acute liver failure, the most common are viral and toxic-medicinal, followed in order of frequency by those that appear during pregnancy and those related to vascular processes. The different causes of acute liver failure are listed below:

Viral. Hepatitis A, B, C, D and E viruses. Other viruses: cytomegalovirus, herpes, Epstein-Barr.

Toxic. Industrial: carbon tetrachloride. Mushrooms: Amanita phalloides.

Medications. Antibiotics and chemotherapy: isoniazid, PAS, tetracycline. Painkillers: paracetamol. Anesthetics: halothane. Other drugs: tricyclic antidepressants, MAO inhibitors, thiouracil, hydantoins, phenothiazines.

Vascular causes. Shock, congestive heart failure, cardiac tamponade, Budd-Chiari syndrome.

Microvesicular steatosis. Severe acute steatosis of pregnancy, Reye syndrome.

Other causes. Wilson's disease, massive tumor invasion of the liver, hyperthermia (heat stroke), generalized bacterial infections (tuberculosis, brucellosis).

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SHORT BOWEL SYNDROME

Synonymy

S. of short intestine malabsorption.

Syndromography

Clinic

It is observed in patients who have had a significant portion of the small intestine resected, who show digestive-absorptive insufficiency that causes malabsorption syndrome. This leads to diarrhea, progressive weight loss, asthenia, loss of muscle mass, lassitude, paleness and symptoms of vitamin-mineral deficiencies.

Paraclinical examinations

Blood. Anemia, hypoproteinemia, low cholesterol, electrolyte alterations.

Stool. Steatorrhea with undigested food remains.

Radiology. It shows the magnitude of the resected segment and the alterations in the remaining segment.

Syndromogenesis and etiology

The disorders that most frequently cause this syndrome are:

1. Extensive intestinal resection due to vascular injury of the small intestine.
2. Multiple intestinal resections due to regional enteritis.
3. Jejunal deviation due to pathological obesity.

In general terms, the absorption of nutrients will depend on the extent and portion of the intestine that was removed, the presence of the ileocecal valve and the adaptation of the remaining intestine. It has been proven that resection of 40-50% of the small intestine is usually well tolerated as long as the duodenum, the distal half of the ileum, and the ileocecal valve are spared. In contrast, resection of only the ileum and ileocecal valve can cause severe diarrhea and malabsorption, even when only 30% of the small intestine is removed.

Literature

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IRRITABLE BOWEL SYNDROME

Synonymy

See irritable bowel syndrome.

Syndromography

This syndrome is the most common gastrointestinal condition in clinics; Although it does not endanger the patient's life, it causes great discomfort to those who suffer from it and creates a feeling of frustration in the doctor who treats them.

This syndrome can take three clinical variants:

1. Patients with so-called spastic colitis or irritable bowel complain mainly of chronic abdominal pain and constipation.
2. Another group has chronic intermittent watery diarrhea that is often painless.
3. Other patients present both characteristics and complain of constipation that alternates with intermittent diarrhea.

Clinic

It is a condition of young adults and its frequency is twice as high in women as in men. The main symptoms are: diarrhea, chronic constipation or both.

The diarrhea is intermittent watery that worsens in the morning when getting up or after breakfast; rarer in the rest of the day or night. They are 3-5 in number with excess mucus. The condition can last weeks or months and disappears spontaneously for a variable period of time before reappearing. Some patients describe "pencil-like" pasty stools instead of diarrhea.

On other occasions the clinical picture is characterized by chronic abdominal pain with constipation that alternates with diarrhea. The pain is colicky in the sigmoid region and is relieved by defecation or passage of gas. Often intense flatulence.

Common are: heartburn, low back pain, lipothymia, palpitations.

They are anxious people. On rectal examination, the ampoule is found empty. Psychological disturbances are observed: depression, hysteria, obsessive-compulsive traits, and any psychological stress usually exacerbates them.

Paraclinical examinations

Colonoscopy. Increased vascularization of the mucosa, difficulty in crossing the rectosigmoid angle, etc.

flexures due to existing spasm. These patients complain a lot during the test.

Colonic manometry. It can detect alterations in the normal basic electrical rhythm that is characterized by increased slow wave activity of three cycles per minute (CPM) in patients with the spastic colon variant.

Syndromogenesis and etiology

The fundamental thing is the alterations of intestinal and/or colonic motility favored by the underlying psychological alterations and which can be triggered by stress. This has been observed in volunteers who have been placed in a stress situation while colonic manometry was performed, which has shown an increase in intracolic pressure.

ISRAEL SYNDROME

Synonymy

S. of hyperbilirubinemia due to shunt.

Syndromography

Clinic

It is observed in both sexes. The beginning is in the second decade of life. It is characterized by jaundice accompanied by splenomegaly.

Paraclinical examinations

Blood. Spherocytes, increased osmotic fragility, moderate reticulocytosis, increased unconjugated bilirubin that persists after splenectomy, autohemolysis, normal mechanical fragility of erythrocytes. Normal liver function tests.

Urine. Increased urobilinogen.

Syndromogenesis and etiology

Israel syndrome is caused by excessive production of bilirubin, directly from the bone marrow from erythrocytes or their precursors, either by destruction or synthesis of hemotetrapyrroles.

Literature

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KÖENIG SYNDROME

Syndromography

Clinic

This syndrome is characterized by diarrhea that alternates with constipation. Recurrent colicky abdominal pain, bloating, borborygmi in the right iliac fossa. A tumor may or may not be palpated in the right lower quadrant.

Paraclinical examinations

Radiology. In the intestinal transit, the lesions produced by chronic infection are observed. Chest radiology shows the lesion due to the primary infection.

Sputum and stool culture. They are important to specify the presence of the tubercle bacillus. Mantoux test: positive.

Syndromogenesis and etiology

It is a tuberculous lesion located at the level of the ileocecal valve, which produces functional disorders at that level and possible subocclusive symptoms.

Literature

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VISCERAL LARVA MIGRANS SYNDROME

Synonymy

S. of tropical eosinophilia. S. of eosinophilia-hepatomegaly.

Syndromography

Clínic

This syndrome is more common in children aged 15-40 months, but also in adults. It generally has a benign course characterized by: low-grade fever, general malaise, anorexia, weight loss, pain in the right upper quadrant. If it is accompanied by respiratory complications such as dyspnea and cough, healing is slow.

The severe form occurs sporadically in isolated cases in a family nucleus and mainly affects children aged 14-40 months. The next oldest sibling usually presents with eosinophilia or another sign of mild or residual infection.

Although for a long time it was considered exceptional in adults, Llanioy Soto were able to present more than 60 cases diagnosed under suspicion of lymphomas or tuberculosis due to the clinical picture, which were accurately diagnosed by laparoscopy.

Symptoms may persist for a year or more, pneumonitis or neurological disorders may occur. Endophthalmitis with loss of vision and a retinal lesion similar to that of retinoblastoma can occur with some frequency in older children, which has led to enucleation of the eye.

The predominant symptoms are itching, cough, nighttime snoring, and febrile convulsions, and physical examination reveals hepatomegaly, pulmonary rales, malnutrition, skin and eye lesions, and lymphadenopathy.

Physical examination. Hepatomegaly, often painful, sometimes discrete splenomegaly.

Paraclinical examinations

Blood. Leukocytosis (which can reach up to 15,000/mm³). Variable eosinophilia always greater than 10-15 in some cases up to 80-90% eosinophils. Hypochromic anemia, hypoalbuminemia, increased IgM. The ELISA technique with larval antigens can be useful in confirming the diagnosis, although it is necessary to exclude other parasites and this test is only performed in highly specialized laboratories.

Chest radiology. Bilateral peribronchial infiltrate. Recurrent in some clinical forms.

Liver biopsy. Normal hepatocytes. Inflammatory reaction consisting of: eosinophils, lymphocytes and plasma cells predominantly. Charcot-Leyden crystals. Sometimes granulomas. The demonstration of ToxOcara larvae confirms the diagnosis, but this procedure is rarely justified.

It is very difficult to obtain larval fragments, even in biopsies directed by laparoscopy.

Laparoscopy. It is the most accurate method to make the diagnosis, by observing elevated, serpentine, yellowish-white lesions on the surface of the liver that represent the traces left by the larvae during their migration.

Syndromogenesis and etiology

Infestation with larvae of Toxocara canis and Toxocara catis.

This syndrome can also be observed in infestations by Fasciola hepatica, Capilaria hepatica and Angioestrogylos, among other parasites.

In short, parasites that do not carry out their normal cycle in man (except Fasciola) and remain in a larval state, invading the tissues: liver, lung, nervous system and eyes, preferably leaving a trace formed by granulomas with eosinophilic predominance in their visceral path.

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INTESTINAL LYMPHANGIECTASIA SYNDROME

Synonymy

S. of hypercatabolic hypoproteinemia. S. of exudative enteropathy. Gordon S.

Syndromography

Clinic

Paraclinical examinations

Blood. Moderate anemia, hypoproteinemia with a decrease in albumin and immunoglobulin G, but also in IgM and IgA, as well as transferrin and ceruloplasmin.

Cholesterol: normal.

There is lymphopenia (due to the loss of lymphocytes with the lymph) and this is accompanied by a delayed hypersensitivity disorder.

Bone marrow. Moderate hyperplasia.

Absorption tests. Steatorrhea that can reach 40 g daily.

Abnormal tolerance to fats and vitamin A. Test for D-xylose and normal sugars. Rapid degradation of radioiodine-labeled albumin with decreased half-life and increased fecal excretion of I-131-labeled polyvinylpyrrolidone.

Radiology. Mucosal edema and image of malabsorption.

Jejunal biopsy. There is no mucosal atrophy. edema of the villi. Dilatation and telangiectasia of lymphatics in the lamina propria and submucosa. Absence of inflammatory reaction.

Syndromogenesis and etiology

Intestinal lymphangiectasia is part of a generalized congenital disorder of the lymphatic system, since there is not only dilation of the intestinal lymphatics but also abnormalities in the thoracic lymphatics (frequently chylothorax), retroperitoneal and peripheral lymphatics.

The mechanism that produces the symptoms, as has been suggested, would be: a hypoplasia of the visceral lymphatic pathways that obstructs or hinders lymph circulation, thereby increasing lymphatic pressure in the intestine. This may cause dilation in the intestinal wall and mesentery.
Steatorrhea and hypoproteinemia are considered to depend on the rupture of dilated lymphatic vessels with lymph leaking into the lumen. There are three facts that support this concept:

1. Chylous fluid has been recovered from the duodenum in these diseases.
mos.
2. The retrograde passage of contrast material from retroperitoneal lymphatics to the duodenum and jejunum has been proven.
3. There may be persistent steatorrhea after instituting a completely fat-free diet, suggesting that there is an increase in intestinal loss of endogenous fat present in the lymph.

It has been proven that the etiology is genetic and is transmitted in an autosomal dominant manner, although the basic defect that causes this mutation is not well defined.

Literature

Cohen, W.N.: “Intestinal lymphagiectasia.” Radiology, 89:1080- -1082, 1967.

Gordon, R.S., Jr.: "Exudative enteropathy. Abnormal permeability of the gastrointestinal tract demonstrable with labeled polyvinylpyrrolidone." Lancet, 1:325-326, 1959.

INTESTINAL LYMPHANGIECTASIA SYNDROME - HYPOBETALIPOPROTEINEMA

Syndromography

Clínica

This syndrome manifests with the same symptoms as intestinal lymphangiectasia, but is accompanied by hypobetaliproteinemia, which is why it is considered a new syndrome.

(See intestinal lymphangiectasia syndrome.)

Literature

Dobbins, W.O.: "Hypo-beta-lipoproteinemia and intestinal lymphangiectasia. A new syndrome of malabsorption and protein-losing enteropathy." Arch. Int. Med., 122:31-38, 1968.

Lewis, L.A. et al.: "Lipid and cytological abnormalities in familial hypo-beta-lipoproteinemia." Abstract. Circulation, 34(suppl.3):19 (Oct.) 1966.

MADIDA SYNDROME

Syndromography

Clinic

This syndrome is characterized by marked hypotonic hypersecretion of tears. It is opposite to sicca syndrome.

Syndromogenesis and etiology

Madida syndrome is assumed to occur as a response of the central nervous system to an injury caused by irritation of the hypothalamus.

Literature

Prinz, C.W.: "Das Madida-Syndrom bei Akrodinie als gegensatz sum Sicca syndrom." Klin. Monatsbl. Augenheilk., 141:749-763, 1932.

POOR DIGESTION SYNDROME

Synonymy

S. of pancreatic insufficiency. Accelerated small intestine syndrome.

Syndromography

Clínica

It is manifested by abdominal distension, flatulence, borborygmi. Occasionally diarrhea.

Paraclinical examinations

Radiology. Decreased intestinal transit time.

Stool. Presence of undigested food (proteins, fats). Decreased or increased duodenal enzymes and lactic acid, depending on the cause. Intolerance to disaccharides.

Syndromogenesis and etiology

It may be due to pancreatic insufficiency, accelerated motility of the small intestine, or disaccharidase deficiency.

Literature

Danhaf, I.E.: "The clinical gas syndromes a pathophysiological approach. " Ann. N.Y. Academic Sci., 150:127-140, 1968.

MALABSORPTION SYNDROME

Syndromography

Clinic

This syndrome occurs in both sexes and at all ages. There are frequently attacks of diarrhea with the expulsion of gray or yellow, soft and greasy stools. Anorexia, weight loss, borborygmi, painful abdominal tension, weakness and fatigue, dyspnea and paresthesia.

Physical examination. Abdominal distention and tympanism, dehydration, hyperkeratosis, folliculitis, hemorrhagic manifestations (ecchymosis, etc.), glossitis, cheilosis.

Paraclinical examinations

They depend on the suspected cause.

Syndromogenesis and etiology

Numerous hereditary or acquired conditions can be the causes of this syndrome. According to the clinical picture and/or paraclinical examinations. The production mechanisms of this syndrome can be considered according to the following topics:

Group I. Inadequate or defective digestion. Postgastrectomy, exocrine pancreatic insufficiency and biliary insufficiency syndromes.

Group II. Metabolic defects. Sprue syndromes, deficiency in the breakdown of sugars.

Group I. Alterations in absorption areas. Resection syndromes, gastroileostomies, fistulas, jejunal exclusion.

Group IV. Conditions of the intestinal wall or lymphatic obstruction. Radiation syndrome, amyloidosis, enteritis (bacterial, viral, parasitic), scleroderma, carcinoid, intestinal cystoid pneumatosis, lymphomas, Whipple syndrome, tuberculosis, Crohn's syndrome, chronic ulcerative jejunitis (non-granulomatous).

Group V. Alterations of the bacterial flora. Blind loop syndromes, multiple jejunal diverticulosis, antibiotic enterocolitis, afferent loop.

Group VI. Endocrinopathies. Diabetes mellitus, hypoparathyroidism, hyperparathyroidism syndromes. Tumor of the islets of Langerhans (Zollinger-Ellison, watery diarrhea-hypokalemia associated with islet adenomas).

Group VIILAprotein normalities. Dysgammaglobulinemia, Bassen-Kornzweig syndromes, intestinal lymphangiectasia, intestinal lymphangiectasia-hypobe-taliproteinemia.

Group VII. Vascular. Syndromes of the superior mesenteric artery, congestive heart failure, Pick, tricuspid regurgitation-protein-losing enteropathy.

Literature

Sleisenger, M. and J. Fordtran: Gastrointestinal diseases. 3" ed. t III. Buenos Aires, Panamericana, 1985.

MALLORY-WEISS SYNDROME

Synonymy

S. of gastroesophageal laceration. S. of gastroesophageal laceration-hemorrhage.

Syndromography

Clínic

This syndrome is more common in men, generally over the age of 30. After a large ingestion of alcohol, a clinical picture begins that is manifested by intense vomiting with strong retching, followed by hematemesis and/or melena.

Physical examination. Pallor, tachycardia, lipotimic state and in some cases shock.

Paraclinical examinations

The studies indicated in hemorrhage should be performed to rule out other conditions.

Esophagoscopy. It should be performed whenever the patient's condition allows. It shows the tear, generally linear, in the juxtacardial region, its extent, and the intensity or cessation of the hemorrhage.

Syndromogenesis and etiology

The hemorrhage is due to a longitudinal tear of the esophageal and gastric mucosa and submucosa in the juxtacardial region.

Literature

Holmes, K.D.: “Mallory-Weiss syndrome: review of 20 cases and literature review.” Ann. Surg. 164:810-820, 1966.

Mallory, G.K. andS. Weiss: "Hemorrhages from lacerations of the cardiac orifice of the stomach due to vomiting." Am. J. Med. Sci., 178:506-5 15, 1929.

MARABLE SYNDROME

Synonymy

S. of celiac compression. S. of the celiac trunk.

Syndromography

Clínica

Marable syndrome is more common in young women.

It is characterized by painful epigastric cramp that begins 30 minutes to 4 hours after ingestion of food, regardless of the type of food. There is moderate relief in the genupectoral position. Sometimes the syndrome is accompanied by nausea, vomiting, belching, flatulence and diarrhea.

Physical examination. Moderate weight loss. Decreased epigastric systolic beat.

Paraclinical examinations

Radiology. Gastrointestinal, does not show alterations.

Aortography. It does not show vascular anomaly.

Syndromogenesis and etiology

This syndrome is due to external compression of the celiac trunk as it passes through the diaphragmatic hiatus. The anterior portion of the hiatus has fibrous tissue that compresses the celiac trunk at its origin. The incision of this tissue makes the symptoms disappear.

Literature

Harjola, PT. and A. Lahtiharju: “Celiac axis syndrome; abdominal angina caused by external compression of the artery". Am.J. Surg., 115:864-869, 1968.

Marable, S.A.; W. Molnar and EM. Berman: "Abdominal pain secondary to celiac axis compression." Am. J. Surg., 111:493-495, 1966.

Marable, S.A.; M.F. Kaplan; F.M. Berman, et al.: Celiac compression syndrome." Am. J. Surg., 115:97-102, 1968.

MEGAESOPHAGUS SYNDROME

Syndromography

Clinic

This clinical syndrome presents with dysphagia, tonic contraction of the cardia, and abnormal dilation of the esophagus.

Syndromogenesis and etiology

Megaesophagus syndrome is seen in patients with severe malnutrition and extensive weight loss. Sometimes it is congenital.

It is frequently caused by schistosomes.

MEGASIGMOID SYNDROME

Syndromography

Clínica

It is observed in elderly people, but also in young people when there is neurological damage. Psychotic reactions They may precede or precede this syndrome. It is observed in patients with neuropsychic symptoms and mental deterioration.

It is characterized by very prolonged constipation masked by anal incontinence of fecal fluid due to loss of sphincter tone. It is accompanied by fever, nausea and vomiting.

Physical examination. Abdominal distension, acute peritonitis or recurrent or intermittent volvulus. Sometimes the liver is somewhat displaced from its usual position.

Paraclinical examinations

Radiology. Irreversible dilation localized to the megasigmoid. Barium enema is impossible.

Syndromogenesis and etiology

Megasigmoid syndrome is always acquired, unlike megacolon, which is congenital in most cases. It is due to motor disorders of the sigmoid of neurological cause in patients with advanced mental deterioration, cerebral arteriosclerosis, Parkinson's disease, multiple sclerosis, tabes.

Literature

Kraft, E. and N. Finby: "Megasigmoid and megasigmoid syndrome." GP., 36:104-114, 1967.

MENETRIER SYNDROME

Synonymy

S. of giant hypertrophic gastritis. S. of hyperprotein gastropathy. S. of gastric adenoma in plaques. S. of localized gastric glandular hypertrophy. S. of tumor hypertrophy of the gastric mucosa. S. of villous gastropathy.

Syndromography

Clínica

This syndrome can occur at all ages, with preference in men who suffer from gastric discomfort.

It is characterized by ulcer-type pain that is relieved with alkalis, anorexia, nausea and vomiting, and weight loss.

Eating food can relieve or suppress pain. Sometimes there is diarrhea, melena and/or hematemesis and steatorrhea.

Physical examination. Generally, it is negative. Sometimes there is a painful sensation on palpation in the epigastrium.

Later, edema and ascites.

Paraclinical examinations

Blood. Hypoproteinemia. anemia, eosinophilia.

Radiology. Marked increase in gastric folds.

Irregularity in the greater curvature. Thin gastric wall.

Mucus mixed with barium.

Gastroscopy. The radiological findings are verified and the extent of the process and the characteristics of the mucosa are specified:
red, with exulcerations, covered in areas by mucus, sometimes signs of recent bleeding.

Gastrochymogram. Hypoacidity.

Biopsy. Taken directly by gastroscopy on any area suspected of malignancy or only to specify the

details. It demonstrates: marked thickening of the folds with hyperplasia and hypertrophy of the mucous glands and loss of almost all the parietal and principal cells. Excess mucus production.

Syndromogenesis and etiology

The etiology is unknown, however inflammatory causes such as cytomegalovirus have been considered; allergic factors, based on eosinophilic infiltration of the mucosa and eosinophilia in the blood. Autoimmune mechanisms are also included, since anti-parietal cell antibodies have been found. Familial involvement is rare, which rules out a hereditary or genetic mechanism.

Hypoproteinemia is due to the loss of plasma proteins through the gastric mucosa, leading to a protein-losing syndrome.

Literature

Butz, W.C.: "Giant hypertrophic gastritis: a report of fourteen cases." Gastroenterology, 39:183-190, 1960.

Chouraqui,J.P.; C.C.Roy, et a.: "Menetrier disease in children. Report of a patient and review of sixteen other cases." Gastroenterology, 80(5): 1042-1047, 1981.

Jeffries, G.H., H.R. Holman and M.H. Sleisenger: "Plasma proteins of the gastrointestinal tract." N. Engl. J. Med., 266:652-660, 1962.

Menetrier, P.: "Des polyadenomes gastriques et des leurs rapports avec le cancer de léstomac." Arch. Physiol. Norm. Path., 1:32-55, 1888.

ABDOMINAL MIGRAINE SYNDROME

Syndromography

Clínica

This syndrome is more common in children. It is characterized by intense and sometimes persistent abdominal pain, accompanied by nausea and vomiting. Sometimes diarrhea. However, a migraine may or may not be present. It must be remembered that in some types of migraine, nausea and vomiting are associated, but they are not accompanied by pain as in this syndrome.

Syndromogenesis and etiology

It is considered a phenomenon equal to migraine and disappears with the same treatment. Crises are more or less frequent.

Literature

Brams, W.A.: "Migraine with pronounced abdominal crisis", linois M.J., 48:244, 1925.

MIKULICZ SYNDROME

Syndromography

Clínica

This syndrome is manifested by dry mouth (xerostomia) and absence of tear secretion.

A chronic bilateral and symmetrical dacryoadenitis is observed, which is also accompanied by inflammation of the salivary glands.

Physical examination. The line of the upper eyelid edge takes on the shape of a stretched italic S. When the eyelid is raised, the gland appears enlarged, edematous, standing out in the superoexternal conjunctival cul-de-sac. Inflammation of the salivary glands, mainly the parotid glands, is also observed.

Bilateral, symmetrical, hard, non-painful swellings of the parotid and lacrimal glands.

Paraclinical examinations

Radiology. Sialography shows dilation of the excretory ducts of the glands in the initial phase and their destruction in the advanced phases. Chest x-ray.

Blood. Hemogram and medullogram (to specify leukemia). Serology.

Sputum, culture and Mantoux test. To specify tuberculosis.

Kweim test. To investigate sarcoidosis.

Syndromogenesis and etiology

It can appear in the following diseases: lymphoid leukemia, lymphomas, sarcoidosis, tuberculosis and syphilis. The most common cause is tuberculosis.

Literature

Casanovas, J.: Ophthalmology. Barcelona, Rocas, 1961. p.l51- -152.

Rubin, P. and B.E. Besse, Jr.: "The sialographic differentiation of Mikulicz's disease and Mikulicz's syndrome." Radiology, 68:477-487, 1957.

Von Mikulicz, J.: Ueber eine eigenartige symetrische Erkankung der Tranen und Mundspeicheldrusen., Beitr. Scream. Forstchr. Gewidmet Theodor Billroth, Stuttgart, 610-630, 1892.

Von Mikulicz, J.: "Concerning a peculiar symmetrical disease of the lacrymal and salivary glands." M. Classics, 2:165-186, 1937.

MILKMAN SYNDROME

Synonymy

S. of pseudofractures. Looser-Milkman S. S. of Osteoporosis-osteomalacia.

Syndromography

Clinic

This syndrome prevails in middle-aged women.

It is characterized by asthenia, pain in the back and legs.

Physical examination. Sensation of pressure in the affected bones. Limp.

Paraclinical examinations

Blood. Low serum calcium and potassium. High alkaline phosphatase.

Radiology. Pseudofractures (Looser or Milkman zones).

Small fissures in the cortex of long bones located symmetrically.

Generalized demineralization.

Syndromogenesis and etiology

It is a disorder of calcium and phosphorus metabolism with osteomalacia. The term Milkman syndrome has been given to indicate the radiological findings (pseudofractures) described by him.

Literature

Milkman, L.A.: “Pseudofractures (hunger osteopathy late rickets, osteomalacia).” Am. J. Roentgen., 24:29-37, 1930.
Steinbach, H.L. and M. Noetzli: "Roentgen appearance of the skeleton in osteomalacia and rickets." Am. J. Roentgen., 91:955-972, 1964.

MINKOWSKY-CHAUFFARD SYNDROME

Synonymy

S. of hereditary spherocytosis. S. of congenital hemolytic anemia. S. of chronic familial jaundice. S. of chronic acholuric jaundice. S. of spherocytic anemia. S. of globular cell anemia.

Syndromography

Clinic

This syndrome occurs at any age and in both sexes. Family history. One of the parents (vesicular dyskinesia) and half of the siblings are affected. Symptoms begin at an early age or adolescence with different degrees of intensity that sometimes do not attract attention:

anorexia, laxity, delayed puberty. Recurrent acute episodes characterized by fever, tachycardia, abdominal pain, dyspnea and vomiting.

Physical examination. Discreet persistent jaundice. Sometimes leg ulcers. Splenomegaly and sometimes discrete hepatomegaly. Anomalies may exist.

Paraclinical examinations

Blood. What is characteristic is the finding of spherocytes in the blood film in greater or lesser quantities.

The most accurate test if there is doubt and to quantitatively assess the presence of spherocytes is oSmotic fragility. As spherocytes have a decreased surface area per unit volume, they undergo lysis more easily when they are exposed to solutions with low salt concentration.

Syndromogenesis and etiology

Although the exact defect in hereditary spherocytosis is not known, it affects cytoskeletal proteins.

Almost all patients have significant spectrin deficiency proportional to the severity of the anemia. The spheroidal contour and rigid structure of the erythrocytes prevent their passage through the spleen; There they are exposed to an environment in which they cannot maintain their intense metabolism. The first injury they suffer to the spleen is a greater loss of conditioning of the membrane surface, which in turn produces a subpopulation of hyperspheroidal erythrocytes in peripheral blood, which will later be destroyed in the spleen.

The molecular pathogenesis of this disorder is not fully defined.

The etiology is due to a mutant gene that is inherited in an autosomal dominant manner, and that expresses the deficiency of ankyrin, a protein necessary to maintain the structure of the cell membrane.

Literature

Chauffard, M.A.: "Pathogenie de l'ictere congenital de 1' adulte." Sem. Med. (Paris) 27:25-29, 1907.
Hanspal, M., et al.: "Molecular basis of spectrin and ankyrin deficiencies in severe hereditary spherocytosis: evidence implicating a primary defect of ankyrin." Blood, 77:165-173, 1991.
Minkowski, O.:"Ueber eine hereditare, unter dem Bilde eines chronischen Ikterus mit Urobilinuric, Splenomegalie und Nierensiderosis velaufende Affection", Verh. Dtsch. Kongr. inn. Med., 18:316-319, 1900.
Wintrobe, M.M.: Clinical Hematology. ed. 6, Philadelphia, Lea & Febiger, 1967.

MIRIZZI SYNDROME

Synonymy

S. of obstruction of the ductus hepaticus. S. of stenosis of the ductus hepaticus.

Syndromography

Clinic

The symptoms are those of hepatocholangiitis (pain, fever, chills, jaundice, pruritus). If the diagnosis is not precise, it can lead to secondary biliary cirrhosis.

Paraclinical examinations

Ultrasound and computed axial tomography. They can make the diagnosis.

Cholangiography. By any route, it requires compression of the hepatic duct and/or obstruction caused by the stone.

Syndromogenesis and etiology

In general, Mirizzi syndrome is caused by a stone impacted in the cystic duct or in the neck of the bladder, which causes partial compression of the hepatic duct because the cystic duct runs parallel to said duct. In some cases, the obstruction may be functionally reflective of the stone phenomenon itself. It may be accompanied by cholecystohepatocholedochal or cystohepatocholedochal fistula.

Bibliography

Albot, G. and M. Cortville: De pathologie van de ductus hepaticus". Belg. t. Geneesk., 10:452-465, 1954.
Mirizzi, PL.: “Hepatic duct syndrome.” G. Int. Chir.. 8:731-777, 1948.

CYSTIC STUMMP SYNDROME

Synonymy

S. of the remaining vesicle. S. of the remaining cystic duct.

Syndromography

Clinic

Symptoms appear several years after a cholecystectomy and are characterized by painful crises in the right hypochondrium with radiation to the shoulder, nausea and vomiting, rarely chills and fever.

Physical examination. Jaundice in most cases.

Positive Murphy maneuver.

Paraclinical examinations

Cholangiography. It shows permeability of the bile duct and the remnant of the cystic duct of greater or lesser extent.

Retrograde cholangiopancreatography. It can show the cystic remnant, with or without stones.

Syndromogenesis and etiology

Formation of a cystic or blind pocket in the region of the cystic duct that contains concentrated bile and may house stones. Other times it is due to neuromas of the cystic stump.

Literature

Brown, M.J.: “Disease cystic duct remnant.” A. M. A. Arch. Surg., 79:304-310, 1959.

Oddi. A.: "Effetti dell estirpazione della cistifellea. Bull. Sc. Med. diBologna, 21:194-202, 1888.

Peterson, F. R.: "Reformed gallbladder review of 27 cases." Tr. West S.A., 51:203-220, 1942.

INTESTINAL CYSTOD PNEUMATOSIS SYNDROME

Synonymy

S. of intestinal emphysema. S. of bullous emphysema of the intestine. S. of peritoneal pneumatosis. S. of cystic lymphopneumatosis. S. of pneumatosis intestinalis. Abdominal cystic gas S. S. de Duvernoy.

Syndromography

Clínic

This syndrome is observed in both sexes and at all ages, although its highest incidence is in males aged between 30-50 years and in the sixth decade in women.

It is characterized by abdominal distension, diarrhea attacks that alternate with constipation. Vague pain. Rectal bleeding.

Partial or total intestinal obstruction is rare.

Physical examination. Crepitation; an abdominal mass is palpated.

Many signs may appear depending on the complications that occur.

Paraclinical examinations

Radiology. Zones of transparent areas that follow the course of the small and large intestine that correspond to multiple gas-filled cysts in the wall of the organ.

They predominate in the splenic flexure and the sigmoid. Occasionally, pneumoperitoneum occurs.

Colonoscopy. Submucosal cystic lesions are observed that are prominent in the lumen according to their size, but the most frequent location is in the small intestine and the cysts can reach up to several centimeters.

Syndromogenesis and etiology

Primary cystic pneumatosis can be associated with other diseases: regional and tuberculous enteritis, ulcerative colitis and others.

The etiology is unknown.

Literature

Bang, B.L.E: "Lufholdige kyster j vaggen af ileum og i nydannet bindevav pammes serosa." Nord. med. ark. (N18), 8:1-15, 1876.

Bockus, H.L.: Gastroenterology. ed. 5, v. III, Philadelphia, Saunders, 1994.

Koss, LG.: “Abdominal gas cysts (pneumatosis cystoides intestine rum hominis)”. Arch. Path., 53:523-549, 1952.

Stone, H.H.; W.B. Allen; R.B. Smith, et al.: "Infantile pneumatosis intestinalis." J. Surg. Res., :301-307, 1968.

INTESTINAL KNOT SYNDROME

Synonymy

S. of double volvulus. S. of complete volvulus.

Syndromography

Clinic

This syndrome is manifested by colicky abdominal pain, distension and ileus that progresses rapidly in cases severe.

Paraclinical examinations

Radiology. Dilated sigmoid colon, located on the right.

Dilated small intestine located on the left side of the abdomen. A contrasting study demonstrates pulsations at the level of the volvulus sigmoid, with an accessory thinning that is located surrounding the ileal loop.

Syndromogenesis and etiology

It is produced by a torsion of the terminal ileum around the sigmoid colon, which undergoes a rotation on its own axis.

Literature

Boyden, FM., et al.: "The intestinal knot syndrome." J.A.M.A. 211:622, 1970.

Kallio, K.E.: "Über volvulus coli transversi." Chir Act. Scand., 70(Suppl. 21):1, 1932.

PROXIMAL JEJUNAL LOOP OBSTRUCTION SYNDROME

Syndromography

Clínica

This syndrome occurs in operated patients who underwent a gastrojejunostomy. Its onset is abrupt with epigastric pain in the left upper quadrant, vomiting without bile, cyclical and recurrent.

Physical examination. Pain on palpation of the epigastrium.

Paraclinical examinations

Radiology and/or endoscopy. They allow the diagnosis of the location of the obstructive phenomenon.

Laparoscopy. It can sometimes determine the extrinsic cause (adhesions), even its lysis during the examination.

Syndromogenesis and etiology

This syndrome is due to an obstruction of the jejunum, after gastric surgery, which may be due to adhesions, gastrojejunal ulcer or failures in the operation.

It is rare today after the development of surgical techniques.

Literature

Bockus, H.L.: Gastroenterology. ed. 5, v. III, Philadelphia, Saunders, 1994.

OGILVIE SYNDROME

Synonymy

S. of acute intestinal pseudo-obstruction. S. of adynamic ileus of the colon.

Syndromography

Clinic

Ogilvie syndrome is characterized by acute intestinal dilation that primarily affects the colon, but sometimes also the small intestine. As in other forms of pseudo-obstruction, clinical signs are difficult to distinguish from mechanical obstruction.

The patient complains of colicky pain in the lower abdomen and acute constipation.

Physical examination. A distended, tympanic abdomen is confirmed, with a decrease or absence of intestinal sounds.

There is often localized hypersensitivity over the distended colon, but it is rare to find: diffuse tenderness in the abdomen, rigidity or rebound hypersensitivity with the sudden decompression maneuver.

Paraclinical examinations

Radiology. The abdomen shows great distention of the colon and small intestine and sometimes air-fluid levels. The cecum, which is the most distensible part of the colon, is usually very dilated and painful.

Colonoscopy. It has been used in differential diagnosis and serves as therapy, by extracting most of the gas and contents of the colon.

Syndromogenesis and etiology

The onset of symptoms is frequently related to illnesses and intense stress, whether medical or surgical, such as recent interventions, myocardial infarction, serious infection, respiratory failure. Frequently, patients have received narcotics or sedatives and suffer metabolic and electrolyte alterations. On other occasions the syndrome occurs without apparent cause.

It is important to clarify that there are no elements that allow us to consider any type of mechanical obstruction.

Literature

Ogilvie, H.: Large intestine colic due to sympathetic deprivation. A new clinical syndrome". Br. Med. J., I: 671-673, 1948.

Zurita Romero, M.; M. Pera; A.Marti Corbella et al.:"Ogilvie syndrome; contribution of 5 cases". Rev. Quir. Esp., 12: 263-267, 1985.

CHRONIC PANCREATITIS SYNDROME

Syndromography

Clinic

This syndrome is more common in males and alcoholics. In females it is associated with biliary diseases. The age of onset depends on the etiology.

It generally begins with an acute pancreatitis crisis or sometimes insidiously and is characterized by flatulence, pain radiating to the left dorsal region; sometimes with other characteristics. Steatorrhea and malnutrition.

If the crises are acute and recurrent, they form another syndrome: Comfort-Steinberg syndrome (see).

Paraclinical examinations

Feces, blood and urine.

Duodenal intubation. It allows the determination of pancreatic enzymes.

Radiology and pancreatography. It shows the alterations of the ducts and calcifications in advanced cases.

Computed axial tomography. It may reveal some useful elements.

Syndromogenesis and etiology

In most cases this syndrome is associated with alcoholism, bile duct diseases (lithiasis and others), viral infections, allergic or hereditary processes.

RECURRENT IDIOPATHIC PANCREATITIS SYNDROME

Syndromography

Clinic

Idiopathic pancreatitis is considered a pancreatitis syndrome in which there is no etiological history demonstrable by conventional methods, for example, absence of viral infection or abdominal trauma, and no medication potentially capable of inducing pancreatitis.

The symptoms are characteristic of pancreatitis.

Syndromogenesis and etiology

The etiology, as its name indicates, is unknown.

PEAN-WERTH SYNDROME

Synonymy

Gelatinous disease of the peritoneum.

Clinic

This syndrome is manifested by a slow-moving and progressive ascites, sometimes preceded by a feeling of discomfort in the abdomen and anorexia. Puncture of the abdomen allows collecting a gelatinous liquid.

Paraclinical examinations

Laparoscopy. Once the liquid has been extracted through a very thick trocar and through the sheath of the laparoscope itself, since it is very thick, air is introduced and a large number of blisters filled with sallow yellow liquid are observed that even hang from the anterior abdominal wall.

Ascitic fluid. Demonstrates the gelatinous substance.

Syndromogenesis and etiology

Pean-Werth syndrome is due to the accumulation of mucoid substance in the peritoneum due to the implantation of muciparous cells from:

1.Appendicular mucoid cyst (Werth's disease).
2. Appendiceal mucocele.
3. Myxocarcinoma of the gastrointestinal tract.
4. Mucinous cyst of the ovary.

PEUTZ-JEGHERS SYNDROME

Synonymy

S. of intestinal polyposis. S. of mucocutaneous pigmentation. S. of melanoplakiaintestinal polyposis. S. of digestive lentiginopolyposis. S. de Peutz. Hutchinson-Weber-Peutz S. S. de Jeghers.

Syndromography

Clinic

This syndrome is observed in both sexes and in all ethnic groups. Symptoms begin in adolescence. It is characterized by severe and recurrent abdominal pain that improves with strong massage maneuvers on the abdomen. Sometimes there is borborygmi and in advanced stages digestive bleeding.

Physical examination. From birth, melanotic pigmentation measuring 2-5 mm in diameter, sometimes coalescing, can be observed on the lips, oral mucosa, palms of the hands and soles of the feet or in the abdominal region. Mucosal pigmentations are permanent but the others can be modified and almost disappear in the course of development.

Paraclinical examinations

Radiology. Intestinal, shows polyps at different levels of the digestive tract.

Blood. Anemia.

Stool. Positive occult blood.

Upper or lower endoscopy. He or she can demonstrate the polyps, including taking samples to classify them and/or determine if they are hamartomas (made up of excess lamina propria and dilated cystic glands).

Malignant degeneration is rare.

Syndromogenesis and etiology

The basic defect of this disease is unknown and although it is of monogenic etiology, it is transmitted by autosomal dominant inheritance with high penetrance.

Literature

Hutchinson, J.: “Pigmentation of the lips and mouth.” Arch. Surg. London, 7:290, 1896.

Jeghers, H.: V.A. McKusick and K.H. Katz: "Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits: syndrome of CLÍNICAI significance". N. Engl. J. Med., 241:993-1005: 1031-1036, 1949.

McAllister, A.J.; N.F. Hicken; R.G. Latimer, et al.: "Seventeen patients with Peutz-Jeghers syndrome four generations." Am. J. Surg.. 114:839-843, 1967.

Peutz, J.L.A.: “Over een zeer merkwaardige, gecombineerde familiaire polyposis van de slimjmvliezen van den tractus intestinalis met die van de neuskeelholte en gepaard met eigenaardige pigemntaties van huiden siijmvliezen. Nedel. maandsxhr. v. geneesk". N.S. 2, 10:134-146, 1921.

PLUMMER-VINSON SYNDROME

Synonymy

S. of sideropenic dysphagia. Paterson-Kelly S. Kelly-Paterson S.

Syndromography

Clinic

It appears in middle age, more common in women. It is manifested by pain and a burning sensation behind the larynx when swallowing solid foods.

Physical examination. Pale skin and mucous membranes. Other peripheral signs of iron deficiency (nails, hair).

Paraclinical examinations

Blood. Hypochromic anemia.

Serum iron. Low.

Syndromogenesis and etiology

It is caused by an iron deficiency. The relationship that may exist between dysphagia and low iron is discussed.

Literature

Plummer, H.S.: *Diffuse dilation of the esophagus without anatomie stenosis (cardiospasm). A report of ninety-one cases'". JA.M.A., 58:2013-2015. 1912.

Vinson, P.P.: “A case of cardiospasm with dilation an angulation of the esophagus." Med. Clin. N. Ame., 3:623-627, 1919.

Vinson, P.P.: "Hysterical dysphagia." Minnesota Med., 5:107- -108, 1922.

Wintrobe, M.M.: "Idiopathic hypochromic anemia." Medicine, 12:187-243, 1933.

FAMILIAR COLON POLYPOSIS SYNDROME

Synonymy

S. of familial adenomatous polyposis of the colon.

Syndromography

Clínica

Polyps appear before the age of 40. Usually around adolescence or shortly after. The symptoms are not specific: sometimes violent, periodic or recurrent diarrhea, sometimes with blood or mucus, weakness and frequently malignant degeneration with the symptoms typical of this condition: change in intestinal habit, obstruction due to intussusception or size of the polyp, symptoms of malnutrition.

Physical examination. Pallor. Sometimes pain over the colon on palpation.

Paraclinical examinations

Radiology. Colon enema shows the polyps, there is integrity of the australis and the wall.

Colonoscopy. It allows viewing and biopsy of suspicious lesions, as well as transendoscopic removal of all possible polyps.

Syndromogenesis and etiology

It is inherited as a high penetrance autosomal dominant family trait, which is present in half of the offspring. The probability of affected individuals suffering from colon cancer approaches 100% at the age of 40, which is why surgery is mandatory (total or subtotal colectomy).

It is considered the most clearly defined precancerous condition in medicine.

Literature

Cripps, H.: “Two cases of disseminated polyps of rectum.” Trans. Med. Soc. London, 33:165, 1882.
Lynch, H.T. and A.J. Krush: "Heredity and adenocarcinoma of the colon." Gastroenterology, 53:5 17-527, 1967.

JUVENILE POLYPOSIS SYNDROME

Synonymy

S. of solitary polyp of colon and rectum. S. of juvenile polyps. Polyp retention S.

Sindomography

Clinic

This syndrome is slightly predominant in young people. It is characterized by recurrent bleeding through the anus with feces.

Sometimes diarrhea, more rarely constipation. When the polyps are large, they can protrude through the anus or cause obstruction or invagination with crampy abdominal pain. They are not accompanied by lesions of other tissues.

Paraclinical examinations

Radiology. Barium enema.

Stool. Positive occult blood.

Colonoscopy. Demonstration of one or more polyps.

Biopsy. It shows a prominent stroma with abundant vascular tissue, infiltration of mononuclear cells and frequent eosinophils. Proliferation of glands, mucous membranes and cysts with mucus. These are hamartomatous polyps.

Syndromogenesis and etiology

The etiology is monogenic and is inherited as an autosomal dominant trait. Members of the patient's family present this type of polyposis and never familial adenomatous polyposis of the colon. This type of polyp does not degenerate.

The basic defect is unknown.

Literature

McKusick, V.A.: "Genetic factors in intestinal polyposis." J.A.M.A., 182:271-277, 1961.
Woolf, C.M.; R.C. Richards and E.J. Gardner: "Occasional discrete polyps of colon and rectum showing inherited tendency in kind." Cancer, 8:403-408, 1955.

POSTCHOLECYSTECTOMY SYNDROME

Synonymy

S. recurrent of the biliary tract.

Syndromography

Clinic

Recurrence of the same symptoms that the patient suffered before cholecystectomy, such as: colicky pain in the right upper quadrant (hepatic colic), distention, nausea and bilious vomiting.

Physical examination. Pain on palpation in the operated area.

Sometimes jaundice.

Paraclinical examinations

Radiology. It is necessary to perform a cholangiography, especially via the transduodenal route, to be able to specify the etiology.

Pancreatic function tests. To rule out associated disease.

Syndromogenesis and etiology

Different mechanisms can explain this syndrome. They can be classified into three groups:

1. Related to surgical intervention:
   Error in the preoperative diagnosis (the symptoms were due to another condition, including extrabiliary).
   There was another concomitant condition (liver, pancreas, neoplasia, stenosis of the sphincter of Oddi, stenosing choledocytitis, cholangiitis).
2. Postoperative consequences:
   - Common bile duct injury or residual stone.
   Intrahepatic stone.
   Remnant cystic duct.
   Adhesions.
3. Functional biliary disorders.

Literature

Burhenne, H.J.: The postcholecystectomy syndrome, in Margulis, A.R. and H.J. (eds.), Alimentary Tract Roentgenology (St. Louis:C. V. Mosby Co., 1973), pp. 1322-1326.
Dreiling, D.A.: “The postcholecystectomy syndrome.” Am. J. Dig. Dis., 7:603, 1962.

POST-DYSENTERIC SYNDROME

Syndromography

Clinic

This syndrome occurs in patients who have had attacks of dysenteric infection, whether amoebic or bacillary, in which the symptoms reappear: pain, diarrhea.

Physical examination. Pain in the right lower quadrant if the infection was due to amoebae, or in the left if it was bacillary dysentery.

Paraclinical examinations

Stool. The persistence of amoebas should be looked for and a culture performed to identify the dysenteric bacillus.

Radiology. The colon may show lesions at that level.

Syndromogenesis and etiology

Postdysenteric syndrome may be due to loss of resistance of the colonic mucosa, irritation of said mucosa, or psychogenic influences. Sometimes a reinfection or an infection that is not completely cured is confirmed.

Literature

Fierst, S.M. and A. Werner: "Postdysenteric syndrome." Gastroenterology, 27:281-29 1, 1954.

POSHEPATITIS SYNDROME

Synonymy

S. of prolonged hepatitis. Convalescent hepatitis S.

Syndromography

Clinic

This syndrome occurs after 56 days of hepatitis; 85% of patients recover completely; 15% present different complications. The symptoms are:

laxity, fatigue, depression, sleep disorders, anorexia, intolerance to fatty foods, weight loss and pain in the upper right quadrant of the abdomen.

Physical examination. Subicterus, slight pain on palpation of the indicated quadrant.

Paraclinical examinations

Blood. All liver tests are normal, except for moderate unconjugated hyperbilirubinemia and slight elevation.

tion of serum cholesterol.

Stool. Mild steatosis.

Liver biopsy. In most cases it is normal;

In 15%, a transition to chronic hepatitis is seen: persistence of the inflammatory reaction and fibrosis.

Syndromogenesis and etiology

Sometimes the cause is psychogenic, other times it is a sequel to acute hepatitis.

Literature

Bockus, HL.: Gastroenterology, ed, 5. v. III, Philadelphia, W.B. Saunders, 1994.

Caravati, C.M.: "Posthepatitis syndrome." South Med. J., 37:251- -257, 1944.

POSTPRANDIAL SYNDROME

Syndromography

Clinic

It is characterized by atypical pain located in the anterior region of the chest that occurs after meals and lasts several hours. There are also palpitations.

Paraclinical examinations

Electrocardiogram. Performed before meals in these patients is totally normal. When performed after meals, during the crisis it shows: sinus tachycardia, depression of the ST segment, flattening of the T wave. If the electrocardiogram is repeated 2-3 h after meals all the changes disappear and are absolutely normal.

In cases that suffer from a previous block, bradycardia is observed.

Syndromogenesis and etiology

The etiology is unknown. (Release of gastrointestinal hormones?)

Literature

Gilbert, N.C.. GK. Fenn and G.V. LeRoy: "The effect of distension of abdominal viscera." J.A.M.A., 115:1962-1967, 1940.

Utsu, F.: V. Enescu; E. Boszormenyi, et al.: "Postprandial syndrome." New Physician, 15:102-103, 1966.

CHRONIC IDIOPATHIC INTESTINAL PSEUDOOBSTRUCTION SYNDROME

Syndromography

Clinic

It is characterized by recurrent attacks of abdominal pain and distension accompanied by nausea and vomiting. Steatorrhea often exists secondary to bacterial overgrowth in the small intestine that can cause chronic diarrhea and malnutrition that frequently lead to death, as the disease tends to be progressive and resistant to pharmacological treatments.

Physical examination. Abdominal distension, tympanism on percussion, absence or notable decrease in fluid sounds, steatorrhea, weight loss and signs of nutritional deficiency.

Paraclinical examinations

Motility. Abnormality of the motility of the small intestine and also the colon. Altered esophageal and bladder motility has been described in many patients.

Radiology. Shows distention with or without air-fluid levels of the intestinal loops and/or colon. Barium examination may find gastric atony and megaduodenum.

Syndromogenesis and etiology

In idiopathic primary pseudo-obstruction no other causal disease can be identified and it is considered a primary disorder of intestinal motility due to alterations in sympathetic innervation or muscular layers. Abnormalities of the myenteric plexus and myopathy of the smooth muscle of the intestine and bladder (the so-called hollow viscus myopathy) have been described. Elevated prostaglandin concentrations have also been reported in some patients.

Two groups of patients have been identified, according to their familial presentation or not and the primary site of the anatomical alterations.

Family type:

1. Familial visceral myopathy type 1, whose etiology is an autosomal dominant gene and familial visceral myopathy type II, whose presentation is due to an autosomal recessive gene.
2. Familial visceral neuropathy, transmitted by an autosomal recessive gene.

Unfamiliar type:

1. Visceral myopathies: a, chronic intestinal pseudoostruction in children, intestinal hypoperistalsis-megabladder-microcolon syndrome; b, visceral neuropathies. Both entities are the only ones that occur in young children and all of them die during childhood due to this condition.

It is worth remembering that there is another similar condition called secondary intestinal pseudoostruction and that, as its name indicates, it is due to demonstrable causes such as:

scleroderma, diabetes, amyloidosis, neuropathies, drugs or infection.

The etiology is monogenic and is transmitted by autosomal recessive inheritance.

Literature

Anuras, S.: “Intestinal pseudoobstruction syndrome.” Ann. Rev. Med., 39:1, 1988.
Schuffler M.D.; AC Rohmann, et al.: "Chronic Intestinal Pseudo-Obstruction. A report of 27 cases and review of the literature." Medicine (Baltimore), 60:173, 1981.

PANCREATIC PSEUDOCYST SYNDROME

Syndromography

Clinic

Symptoms begin after the patient has suffered severe abdominal trauma, attacks of acute pancreatitis or gallbladder disease, and are characterized by pain of medium to severe intensity, generally located on the right side of the epigastrium or in the left upper quadrant, accompanied by nausea and vomiting that suggest a paralytic ileus or pyloric obstruction. Also, progressive weight loss.

Physical examination. A tumor is palpated in the epigastrium or upper lateral quadrant, 10-15 cm in diameter, non-displaceable, with fever and discomfort in that area. Occasionally paralytic ileus and less frequently, left hydrothorax and/or jaundice.

Paraclinical examinations

Radiology. Simple abdomen. Stomach and duodenum with barium, bile ducts (cholangiography). and thorax.

Ultrasound and computed axial tomography. To locate the cyst and its size, these tests are very useful, with or without contrast, in these cases.

Blood. Serum amylase double normal. Leukocytosis in half of the cases.

Syndromogenesis and etiology

This pseudocyst is a consequence of abdominal trauma or attacks of acute pancreatitis. The symptoms depend on the compression on neighboring organs, especially the digestive tract, and/or irritation of the pancreatic tissue surrounding the cyst.

Literature

Gussenbayer, C.: "Zur operativen Behandlung der Pankreas-Cysten" Arch. Klin. Cir., 29:358-364, 1883.
Wirsung, T.S. and L.B. Costopoulos: "The diagnosis and treatment of pancreatic pseudocysts". Can. Med. Ass. J., 97:1117-1128, 1967.

PSEUDOULCEROUS SYNDROME

Synonymy

Non-ulcer dyspepsia.

Syndromography

Clínic

This syndrome groups diseases and/or processes that present with symptoms that make up a clinical picture that is reminiscent of and can be confused with gastroduodenal peptic ulcer.

Epigastric abdominal pain is the main symptom, it can be located in the upper or middle epigastrium, depending on the cause, it is not usually relieved by the ingestion of food or antacids as occurs with the so-called epigastric hunger pain, corresponding to most duodenal ulcers. The nature of the pain can be burning or stinging, aching or dull, and in the majority of patients it is of medium or low intensity. Irradiation is variable and can be upward, to the right or to the left, also depending on the causal disease. Although the epigastralgia caused by this syndrome does not have a typical periodicity as corresponds to that produced by gastric and duodenal ulcers, some gastritis and duodenitis can sometimes present with periodic epigastric pain. Belching, gastric burning, gastric fullness, slow digestion, flatulence, and sometimes regurgitation are typical symptoms of this syndrome.

Paraclinical examinations

Contrasted study of the stomach and duoene. It must be performed for the diagnosis of hiatal hernia.

Biliary drainage. In search of the parasites that cause the syndrome.

Upper digestive endoscopy. It will show the characteristic images of esophagitis, gastritis, duodenitis and hiatal hernia. Through this procedure, samples can be taken to perform smears of the duodenal mucosa in search of parasites (mainly giardia) and an antral mucosa sample for diagnosis of Helicobacterpylori.

Syndromogenesis and etiology

Hiatal hernia and its fundamental complication, reflux esophagitis, can be the cause of this syndrome.

Gastritis and duodenitis typically present with a pseudoulcer syndrome. Parasitic duodenitis due to Giardia lamblia, necator and strongyloides constitute one of the etiologies of this syndrome.

Helicobacter pylori is associated with pseudoulcer syndrome through its etiological relationship in chronic gastritis and is currently one of the most frequent causes of this syndrome, although the processes mentioned above must be ruled out. It can coexist with some of the parasitic infections mentioned.

Literature

Bockus, H.: Gastroenterology. ed. 5, v. I. 1996.
Llanio, R.: Clinical propedeutics and pathophysiology. People and Education, Havana, 1981, pp:513-528-532.

HEPATIC CYST SYNDROME

Synonymy

S. of congenital hepatic fibrosis.

Syndromography

There are two forms depending on age: child-adolescent type, adult form.

The child-adolescent type has the following characteristics:

Clínica

It affects both sexes equally. It presents with digestive bleeding caused by esophageal varices. It is accompanied by splenomegaly.

Paraclinical examinations

Blood. Normal or slightly elevated bilirubin. Elevated alkaline phosphatase. Elevated thymol clouding, as well as oxalacetic and pyruvic transaminases. Moderately decreased prothrombin time. Normal excretion of bromosulfthalein.

Liver biopsy. Proliferation of connective tissue in the periportal region. Greater or lesser dilation of the bile ducts. Lobular architecture and preserved liver cells.

Descending urography. Normal or with a polycystic pattern (see renal cystic syndrome). Increase in portal pressure.

Syndromogenesis and etiology

It appears to be a variant of polycystic disease.

In this disease, cysts coexist in other organs: kidney, ovary, lung.

The etiology is unknown.

The adult type has the following characteristics:

Clinic

It is characterized by dyspepsia, abdominal pain, and hemorrhages due to rupture of esophageal varices.

Physical examination. Massive hepatomegaly, splenomegaly.

Paraclinical examinations

They give the same results as in the child-adolescent type.

Syndromogenesis and etiology

Same as the previous type.

Literature

Clermont, R.J.; J.N. Maillard; J.P. Benhamon, et al.: "Congenital hepatic fibrosis." Dog. Med. Assoc. J., 97:1272-1278, 1967.

Wilcox, R.G. and K.J. Isselbacker: "Chronic liver disease in young people." Am. J. Med., 30:185-195, 1961.

RAPUNZEL SYNDROME

Synonymy

S. of ileus due to trichobezoar.

Syndromography

Clínica

The name of the syndrome was given due to its resemblance to the character in Grim's fable, who had very long hair. It occurs in psychopathic patients who constantly ingest their hair.

History of tendency to ingest foreign bodies.

It begins with intermittent abdominal pain of progressively intense colicky type, tachycardia, fever.

Physical examination. Palpable tumor with resistance to palpation, marked borborygmi.

Paraclinical examinations

Radiology. Air-fluid levels in the upper part of the small intestine. Contrast arrest in the small intestine.

Syndromogenesis and etiology

The mass of hair migrates from the stomach to the intestine and remains imprisoned, advancing slowly through peristaltic movements, until it is impossible to advance, and this gives rise to mechanical ileus.

Literature

Vaughan, E.D.; J.L. Sawyers and H.W. Scot: "The Rapunzel syndrome: An unusual complication of intestinal bezoar." Surgery, 63:339-343, 1968.

RIGHT RECTAL SYNDROME

Synonymy

S. pseudoappendicular.

Syndromography

Clínica

Intense pain located, at the beginning, in the lower right quadrant of the abdomen. Nausea without accompanying vomiting (absence of generalized cramps to the rest of the abdomen).

Physical examination. On palpation there is discomfort and contracture at McBurney's point. No pain upon sudden decompression.

Persistence of painful discomfort when the rectus muscle is ordered to contract, which can be reproduced if the patient raises the neck or tries to sit without hand support.

Symptoms of rheumatic syndrome may be present.

Paraclinical examinations

Blood. In rheumatic fever there is elevated erythrocyte sedimentation rate, moderate leukocytosis, and the differential leukocyte count is normal. Antistreptolysin titer, normal.

In cases where there is a muscle injury, blood tests are normal.

Laparoscopy. It is a decisive method in doubtful cases, since it allows the normality of the appendix and other abdominal structures to be verified.

Syndromogenesis and etiology

Right rectus syndrome is due to rheumatic myositis with damage to the right rectus abdominis.

Literature

Gettill, P.: “Differential diagnosis of abdominal manifestation of acute rheumatic fever from acute appendicitis.” Ann.Surg., 99:650-660, 1934.

Reitman, N.: “Abdominal manifestation of rheumatic fever: description of a right rectus syndrome". Ann. Intern. Med., 22:671-687, 1945.

STEAK RESTAURANT SYNDROME

Synonymy

S. of the lower esophageal ring.

Syndromography

Clinic

Symptoms of incomplete obstruction of the esophagus occur, often due to eating food too quickly and in

especially if they are pieces of meat that have not been properly chewed. It is characterized by acute dysphagia. It is good to remember that there is a clinical picture that also occurs in a meat restaurant, but in this case the piece of meat stops between the pharynx and the epiglottis just before passing into the esophagus, which causes the closure of the larynx and produces signs of asphyxiation. In these cases, the Heimlich maneuver, taking advantage of the residual air left in the lung, can save the person's life, otherwise they can die in minutes.

Paraclinical examinations

Radiology. Smooth esophagus, concentric ring in the distal portion of the esophagus that is not easy to see on routine fluoroscopy and must be demonstrated with the esophagus well dilated by a thick barium mixture.

Esophagoscopy. It is difficult to prove by this method.

Syndromogenesis and etiology

It is due to a passive fibrous structure located at the esophagogastric junction, almost always accompanied by a hiatal hernia.

Literature

Norton, R.A. and G.D. King: "Steakhouse syndrome; the symptomatic lower esophageal ring." Lahey, Clin. Bull., 13:55-59, 1963.

Schatzki, R. and J.E. Gary: "Dysphagya due to diaphragm-like localized narrowing in the lower esophagus (lower esophageal ring)." Amer. J. Roentgenol., 70:911-922, 1953.

CHINESE RESTAURANT SYNDROME

Synonymy

Chinese headache syndrome.

Syndromography

Clínica

Symptoms begin 10-20 minutes after eating the first food in a Chinese restaurant (especially Won Ton soup), and are manifested by stiffness of the facial muscles and neck pain, radiating pain towards the arm with a sensation of paralysis. General adynamia. Distressing cardiac sensation similar to a heart attack. Temporary headache, hot flashes and 1-2 minutes later, profuse sweating.

Syndromogenesis and etiology

This syndrome is observed in people who eat in some Chinese, Japanese or Polynesian restaurants. It is considered that it may be due to the monosodium l-glutamate that comes in some foods. This substance induces a temporary hyponatremia that causes a decrease in intracellular potassium.
The possibility has also been considered that tannin or certain tea ferments or another toxin contained in certain fish (tetro-dotoxin) could be the cause of the clinical picture.

Literature

Ho Man Kwok, R.: "Chinese restaurant syndrome". N. Engl. J. Med., 278:796, 1968.

Lehrer, L.: "A questionnaire study of the prevalence of Chinese restaurant syndrome". Fed. Proc., 36: 1617-1623, 1977.

KINGE'S SYNDROME

Synonymy

Massive fatty liver disease with acute encephalopathy.

Syndromography

Clinic

This syndrome occurs almost exclusively in children under 15 years of age, frequently between 6 months and 10 years: in both sexes. The condition usually begins after an upper respiratory infection, mainly influenza or chickenpox. Within 2-3 days, vomiting occurs followed by stupor that usually progresses rapidly to generalized seizures and coma (signs of damage to the central nervous system).

Physical examination. Hepatomegaly is detected but it is very significant that there is no or minimal jaundice. Some patients adopt characteristic positions in bed.

Paraclinical examinations

Blood. Increased aminotransferases, prolonged prothrombin time, hypoglycemia, metabolic acidosis, and increased serum ammonia. Negative viral hepatitis markers. Leukocytosis with neutrophilia.

Cerebrospinal fluid. Hypoglycorrhachia.

Liver biopsy. It should be done with great caution and according to the prothrombin time. It is observed:
Extensive fatty vacuolization of liver cells. The electron microscope reveals alterations in the mitochondria.

Syndromogenesis and etiology

The cause is unknown, it is attributed to different antiviral and toxic agents, especially salicylates. In many cases there are no medications as antecedents. Mortality is around 50%, but in the cases that survive, chronic liver injury is not demonstrated.

Literature

Brain, W.R. and D. Hunter: "Acute meningo-encephalomyelitis of childhood, report of six cases." Lancet, 1:221-227, 1929.

Laxdal, O.E.; R.P. Shinha; J. Merida, et al.: "Reye's syndrome. Encephalopathy in children associated with fatty changes in the viscera". Amer. J. Dis. Child., 117:717-721. 1969.

Reye, R.D.K.; G. Morgan and J. Baral: "Encephalopathy and fatty degeneration of the viscera. A disease entity in childhood." Lancet, 2:749-752, 1963.

ROTOR SYNDROME

Synonymy

S. of idiopathic hyperbilirubinemia type Rotor.
Hyperbilirubinemia type I.

Syndromography

Clinic

It is manifested by jaundice of the skin and mucous membranes, sometimes subicterus without any other manifestations.

Physical examination. No hepatomegaly or splenomegaly.

Paraclinical examinations

Blood. Direct hyperbilirubinemia. Normal liver tests if certain delays in the elimination of bromosulfthalein, rose bengal and radiological contrast are not taken into account.
Hypercholesterolemia is not rare. There are no signs of hemolysis.

Urine. Bilirubinuria.

Cholecystography. Vesicle excluded.

Liver biopsy. Normal.There is no pigment deposit in the liver cells. Under the electron microscope, numerous giant mitochondria with inclusions of a reticular structure and the presence of microvilli can be observed in the hepatocyte.

Syndromogenesis and etiology

Probable hereditary familial liver disorder in the elimination of bilirubin, bromosufthalein, rose bengal and radiopaque media.

Hereditary transmission is autosomal recessive.

Literature

Rotor, A.B.; L. Manahan and A. Florentin: "Familial nonhemolytic jaundice with direct van den Bergh reaction." Acta Med. Philipp., 5:37-39, 1948.

Wolkoff, A.W.; E. Wolpert, et al. "Rotor's syndrome. A distinct inheritable pathophysiological entity." Am. J. Med., 60:173-179, 1976.

ROSENBACH SYNDROME

Syndromography

Clinic

This syndrome begins in the third or fourth decade of life and is manifested by a sensation of paroxysmal general anxiety with precordial angina, palpitations, arrhythmia, dyspnea, and tightness in the epigastrium. Pulsations can be palpated in the epigastrium that are sometimes visible, there is prominence of that region. Paroxysmal bulimia difficult to control. The patient maintains good general condition, although he normally eats little, especially fruits, cold foods, and cold water.

Syndromogenesis and etiology

The etiology is not well known, it is considered that it may be due to a reflex excitation of the vagus by action on the gastric branches, or that it may be an intestinal allergy with secondary vagal neurosis.

SAINT'S SYNDROME

Synonymy

S. of Saint's triad (diaphragmatic hernia-cholelithiasis-diverticulosis of the colon)

Syndromography

Clinic

The symptoms are related to the triad. Symptoms of hiatal hernia and those specific to cholelithiasis and colonic diverticulosis. However, the syndrome can be asymptomatic for a long time.

Syndromogenesis and etiology

It is debated whether it is a true syndrome due to a flaccidity of the connective tissue that causes hiatal hernia and diverticula with a tendency to gallstones; or a casual association, since cholelithiasis is very common.

Literature

Palmer. E.D.: "Further experiences with Sint's triad: hiatus hernia gall stones and diverticulosis coli." Am. J. Med. Sci., 244:70, 1962.

SCHÖNLEIN-HENOCH SYNDROME

Synonymy

S. of Henoch-Schönlein. S. of anaphylactoid purpura. S. of allergic purpura. S. of rheumatic peliosis. S. of hemorrhagic capillary toxicosis.

Syndromography

Clínica

This syndrome is more common in children and young adults.
Rarely in elderly people. It predominates in the male sex. The onset is variable: headache, anorexia, moderate and irregular fever, abdominal and/or joint pain.

Sometimes skin manifestations occur such as a maculopapular rash due to vasculitis that can also affect joints and kidneys, sometimes there is hematuria. Abdominal pain initiates the syndrome in 35-60% of cases, it is colicky and almost always occurs at night. Sometimes it can be accompanied by vomiting, diarrhea with mucus and blood or constipation.

The syndrome has a tendency to regress and reappear several times in weeks or months and other times ends by spontaneous resolution.

In rare cases it can evolve into a chronic form.

Physical examination. There is no abdominal contracture. Rheumatic pain can affect the joints, mainly in the legs, with periarticular effusions. The most frequent skin manifestations are: purpura, purplish urticaria, angioneurotic edema, diffuse erythema, necrotic ulcer, especially on the legs, rarely in the oral cavity.

The nervous system and other organs may be affected:
paresis, seizures, optic atrophy, ophthalmitis.

Paraclinical examinations

Blood. Normal or very slight platelet count diminished you. Minimal leukocytosis with or without eosinophilia.
Cryoglobulinemia may occur. Positive rheumatoid factor.
Accelerated erythrocyte sedimentation.

Radiology. At first it may reveal the intestinal edge with a punctate appearance and later finger-shaped alterations or slough of the mucosa. These alterations disappear when the patient recovers.

Laparoscopy. It is decisive to determine the abdominal condition. It shows intact organs and at the same time characteristic petechial suffusions in several intestinal loops that perfectly explain the clinical picture.

Determination of immune complexes.

Urine. There is hematuria and blood casts.

Loop test. Positive.

Syndromogenesis and etiology

It can be triggered by various antigens, such as: infectious agents, drugs, some foods, insect bites, immunizations with the formation of immunocomplexes. Hypersensitivity reaction (hypersensitivity vasculitis).

The painting can be repeated as long as they occur similar conditions.

Literature

Henoch, E.: "Über eine eigenthumliche Form von Purpura." Berl. klin. Wschr. 11:641-643, 1874.

Rodríguez-Erdmann, F. and R. Levitan: “Gastrointestinal and roentgenological manifestations of Schöenlein-Henoch purpura.” Gastroenterology, 54:260-264, 1968.

Schönlein, J.L.: “Allgemeine und specielle pathologie und Therapie. Nach dessen Vorlesungen nmiedergeschrieben und hrsg. Von inigen seiner Zuhö". 3. aufl., Herisou, Lit-Comp., 1837.

ACIDOGASTRIC SECRETION SYNDROME I

Syndromography

Clinic

Belching, gastric distention, flatulence.

Paraclinical examinations

Tests for gastric acidity and gastric and duodenal bacterial flora should be performed.

Syndromogenesis and etiology

This syndrome is caused by gastric hypoacidity that must be therapeutically corrected to prevent bacterial growth.

ACIDOGASTRIC SECRETION SYNDROME I

Syndromography

Clínica

It is characterized by epigastric pain, abdominal distension and flatulence.

Paraclinical examinations

The tests indicated in the previous syndrome must be performed and stools must also be taken to determine or rule out occult blood and/or mucus.

Syndromogenesis and etiology

It is due to gastric hyperacidity (with or without peptic ulcer) that is associated with the production of carbon dioxide.

Literature

Danhaf, ILE.: "The clinical gas syndromes, a pathophysiological approach." Ann. N.Y. Acad. Sci., 150:127-140, 1968.

SILVESTRINI-CORDA SYNDROME

Synonymy

of liver cirrhosis-endocrine deficiency.

Syndromography

Clinic

This syndrome is characterized by the association of atrophic cirosis with symptoms of endocrine deficiency. These symptoms differ in both sexes.

Male sex:

1. Loss of armpit and body hair.
2. Impotence, decreased libido.
3. Gynecomastia.
4. Spider veins.

Female sex:

1. Alterations of the menstrual cycle.
2. Sterility.
3. Characteristic atrophic changes due to the absence of ovarian function.
4. Lack of estrogenic activity.
5. Decreased excretion of androgens, 17-ketosteroids and follicle-stimulating hormone.

Furthermore, all the characteristic symptoms of cirrhosis in the advanced stage can be observed in both sexes.

Syndromogenesis and etiology

The endocrine symptoms are explained by the deficiency in hormone production and/or the lack of inactivation of hormones by injured liver cells. The excess of unconjugated estrogenic hormones in the liver allows their accumulation in the blood and this inhibits the follicle-stimulating hormone of the pituitary gland, which of course causes the symptoms in women.

STUART-BRAS SYNDROME

Synonymy

S. of obstructive phlebitis of the small intrahepatic veins.

Syndromography

Clinic

This syndrome is seen mainly in Western India, preferably in children.

It is manifested by acute pain in the liver region with ballooning of the abdomen. Hepatomegaly, ascites, absence of jaundice

The evolution is variable, some cases improve after 4-6 weeks and the biochemical alterations normalize. In other cases the clinical picture ends in liver failure and death or in liver cirrhosis.

Paraclinical examinations

Liver biopsy. Centrilobular edema with blood lakes, dilated sinusoids, cellular necrosis, subendothelial edema and/or proliferation of the reticulum, obstruction of the venules.

Blood. Alterations in liver function tests, hypergammaglobulinemia and alterations in colloidal lability tests.

Syndromogenesis and etiology

It is considered very likely that it is due to toxic damage from different plant substances. It can be caused experimentally in the animal by poisoning with extracts of crotalaria and senecio.

Literature

Bras, G.: D.B. Jeliffe and K.L. Stuart: “Veno-acclusive disease of the liver with nonportal type of cirrhosis, occurring in Jamaica.” Arch. Path., 57:285-300, 1954.

Bras, G.; D.M. Berry and P. Györghy: "Plants as aetiological factor in veno-occlusive disease of the liver" Lancet, 1:960-962, 1957.

TURCOT SYNDROME

Synonymy

S. of glioma-polyposis.

Syndromography

Clínic

The symptoms are a sum of those observed in colon polyposis (see familial polyposis) and those of a tumor of the central nervous system.

It begins at 3-6 years of age due to digestive bleeding in feces, mixed with mucus without being accompanied by pain.

Other times blood occurs at the end of the bowel movement.

Sometimes a polyp prolapses. Tumor, brain or spinal symptoms begin after the age of 25.

Paraclinical examinations

The examinations are those used in the two conditions mentioned here.

Syndromogenesis and etiology

It is a hereditary condition that is transmitted on an autosomal recessive basis. It has only been reported in two brothers born from a consanguineous marriage. The lesions are polyps with a tendency to malignancy. Tumors of the central nervous system are glioblastomas, astrocytomas and medulloblastomas.

Literature

Turcot, J.; JP. Despres and F. Pierre: “Malignant tumor of the central nervous system associated with familial polyposis of the colon: report of two cases.” Dis. Colon Rectum, 2:465, 1959.

CURLING ULCER SYNDROME

Syndromography

Clínic

Repeated and profuse rectal bleeding that occurs after a severe burn. Gastrointestinal perforation with peritonitis may occur. Massive gastrointestinal bleeding during a burn is rare (2-4%); However, moderate bleeding is common (58%).

Paraclinical examinations

Panendoscopy. It allows diagnosing the ulcer, especially if it is located in the upper digestive tract, determining its characters and whether it is bleeding or not, you can even perform a therapeutic action locally and try to stop the bleeding.

Syndromogenesis and etiology

Curling's ulcer represents the final stage of small gastrointestinal erosions that fuse, enlarge and deepen, injuring the submucosal vessels. It is common in severe burns.

The etiology is unknown

Literature

Curling., T.B.: “On acute ulceration of the duodenum in cases of burn." Medico-Chir. Trans., London, 25:260-281, 1842.

Ousterhout, D.K. and I. Feller: "Occult gastrointestinal hemorrhages in burned patients." Arch. Surg., 96:420-422, 1968.

GASTRODUODENAL ULCER SYNDROME - CHRONIC LUNG DISEASE

Synonymy

S. of chronic lung disease-gastroduodenal ulcer.

Syndromography

Clinic

It is known that gastroduodenal ulcer in the general population is in the range of 10%. However, in patients with chronic lung diseases, the incidence is 25%, which is statistically significant and therefore this association has been considered a syndrome.

Syndromogenesis and etiology

The etiology is unknown, although it is considered that the hypoxia factor caused by chronic lung disease may be involved.

Literature

Green, T.P. and V.C. Dundee: *On the association of chronic pulmonary emphysema with chronic peptic ulceration". Can. Med. Assoc. J., 67:438-439, 1952.

West, W. O.; R.O. Burns; J.M. Daniel, et al.: "The syndrome of chronic pulmonary disease and gastroduodenal ulceration." Arch. Intern. Med., 103:897-901, 1959.

ULCERUS SYNDROME

Synonymy

S. of gastroduodenal peptic ulcer. Cruvelhier's ulcer. Ulcer disease.

Syndromography

Clínic

Pain in the epigastrium is the most common symptom in gastroduodenal peptic ulcer; it generally does not radiate, although in certain ulcers of the small posterior contact curvature and some duodenal ulcers it can radiate backwards. The character of the pain can vary, from being dull, burning or burning, in the form of a cramp.

or twisting to truly terrifying pain, although the latter is usually observed in perforating ulcers. The intensity of the pain also varies, from very intense to less intense forms. Relief is achieved with the ingestion of food or antacids.

The schedule rhythm of ulcer pain has been described; that of gastric ulcer is called the "four-beat rhythm" (food, relief of pain, early reappearance of pain, new relief of pain). The rhythm of duodenal ulcer is called “three-beat rhythm” (food, relief of pain, delayed recurrence of pain). Although currently this rhythm or schedule is accepted only as a guide.

The pain is of typical periodicity. The painful periods last 2 or 3 weeks, or less frequently 4 and up to 2 months in exceptional cases. Interspersed periods of calm or remission are observed, so clear that the patient believes himself completely cured.

The pain of gastric peptic ulcer is sometimes accompanied by vomiting, which is rare in duodenal ulcer.

The pain characteristics described are typical of uncomplicated peptic ulcer disease. Some patients may never experience pain and are diagnosed with an ulcer only after a complication has arisen. The appearance of the latter can cause the characteristics of the pain to change.

The physical examination of the patient provides little information. Palpation of the epigastrium produces pain that is often not intense.

Paraclinical examinations

Stomach and duodenum radiology. The typical benign gastric ulcer appears along the lesser curvature and is usually seen in profile position as the addition of a line protruding from the barium-filled stomach. In this location the ulcer crater projects beyond the lumen of the stomach and will appear as a smooth, conical or button-shaped "niche", whose edges are rounded and well defined. Other important diagnostic features include the gastric folds surrounding the ulcer; these must remain intact and extend uninterrupted to the edge of the crater.

In duodenal ulcer, a "niche" or projection of barium is observed in profile or as a spot or collection of barium from the front. More than 90% are located in the proximal two-thirds of the bulb.

Endoscopy. This technique has increased diagnostic precision. The gastric ulcer is characterized by having well-defined edges with a clean yellowish-white background, the folds are directed towards the crater without deformities.

The duodenal ulcer manifests itself as a whitish or yellowish crater because it is covered with fibrinoid necrosis. The usual size varies between 5 and 15 mm. In general, they are single but can be multiple. The morphological appearance varies due to the different evolutionary stages; The most common shape is oval and rounded, and is usually surrounded by a reddened inflammatory ring that corresponds to the acute phase of the injury.

Syndromogenesis and etiology

Peptic ulcer is considered to develop due to an imbalance between the secretion of acid and pepsin and the resistance of the gastric or duodenal mucosa.

In duodenal ulcer the fundamental cause lies in the gastric acid hypersecretion. However, in patients with gastric ulcer, defective mucosal resistance seems to be the main factor.

Overall, patients with duodenal ulcer secrete more acid than normal. Those who suffer from it have a greater mass of parietal cells than individuals without duodenal ulcer, they also have an increase in gastric secretion of pepsin and serum concentrations of pepsinogen I.

Fasting serum gastrin concentrations are normal in patients with duodenal ulcer; however, these patients have a greater acid secretory response to gastrin than normal people. Additionally, intragastric acid is less effective in inhibiting gastrin release and acid secretion.

Patients with duodenal ulcer tend to empty their stomach more quickly than others. This phenomenon, combined with acid hypersecretion, perhaps contributes to the high concentrations of hydrogen ions in the first portion of the duodenum that characterizes these patients.

A significantly lower frequency of retrograde motor sequences has been described in the duodenum of patients with duodenal ulcer. This anomaly could alter the retrograde transport of bicarbonate to neutralize the acid in the bulb.

The possibility of less efficient motility in the duodenum has also been suggested, allowing the acid to remain in contact with the duodenal mucosa for longer.

Genetic factors are important. Duodenal ulcers are three times more common in first-degree relatives of patients with duodenal ulcer when compared to the general population. These patients form a greater proportion of O blood groups and non-reactor status, they do not secrete AB(H) antigen in their gastric juice. A higher frequency of HLA-B5 antigen has also been observed in white men with duodenal ulcer. Increases in serum pepsinogen I appear to be an autosomal dominant character. Individuals with this character have a frequency of duodenal ulcer eight times higher than the general population.

Although the psychological factors in the pathogenesis of duodenal ulcer continue to be the subject of discussion, everything seems to indicate that chronic anxiety and psychological stress are factors that exacerbate ulcer activity.

The relationship between cigarette smoking and a greater frequency of duodenal ulcer, as well as greater motility, is known; perhaps this is due to the inhibition of pancreatic bicarbonate secretion, acceleration of gastric emptying, or both together.

In the pathogenesis of gastric ulcer, almost all data point to the importance of primary defects in mucosal resistance, direct injury, or both as the most important elements. Various factors have been proposed as possible participants in mucosal resistance.

Gastric mucus secreted by mucosal cells has been suggested to be involved in the protection of the mucosa, preventing peptic

The gastric mucosal barrier for retrograde diffusion of hydrogen ions constitutes mucosal resistance to acid-peptic ulceration. This barrier can be destroyed by bile acids, salicylate, alcohol and weak organic acids.

It has been proven that patients with gastric ulcer have a greater reflux of bile into the stomach when compared to non-ulcer individuals or individuals who suffer from duodenal ulcer.

This is assumed to be due to a defect in the function of the pyloric sphincter.

Bile acids contained in bile damage the mucosa gastric by destroying its protective mucosal barrier and thus promoting the retrograde diffusion of hydrogen ions, causing cellular damage, release of histamine by mast cells, more stimulation of acid secretion, destruction of small blood vessels, ulceration and mucosal hemorrhage.

The maintenance of normal blood circulation in the mucosa is an essential factor in the resistance of the mucosa to injury.

Several prostaglandins, especially those of the E series, have been shown to inhibit mucosal injury and therefore have a cytoprotective function. Injury or slight irritation of the mucosa can induce the synthesis of prostaglandins and thus possibly reinforce the resistance of the mucosa, a concept known as adaptive cytoprotection.

The causal relationship that exists between Helicobacter pylori and gastroduodenal peptic ulcer is recognized, having an important role in its pathogenic nechanism.

The frequency of this bacteria in histologically normal mucosa is very low, less than 10% with extreme figures between 2.9 and 29%. In gastric ulcer the percentages of its discovery fluctuate between 70 and 92% and in duodenal ulcer from 80 to 95%. It has been detected in up to 100% in recurrent duodenal ulcer.

Helicobacter pylori has a high affinity for the gastric epithelium and has virtually never been observed on any other surface, however it has been found outside the stomach, in islets of gastric metaplasia in the duodenum and esophagus.

An inflammatory reaction occurs in response to H. pylori infection, which stays tightly adhered to gastric epithelial cells, below the protective layer of mucus.

The presence of the bacteria is associated with cellular changes. There is loss of intercellular junctions between the mucous cells, which curve and show loss of uniformity of the cell surface, there is a decrease in microvilli and loss of the cytoskeleton at their base. The mucus contained in the mucosal cells is generally reduced, with intracellular edema and the presence of phagolysosomes observed.

Cellular alterations progress and microvascular injury occurs, leading to damage that leads to chronic gastritis, damage that can progress and lead to gastric atrophy. Gastritis precedes the formation of peptic ulcer.

Literature

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Goodwin, C.S.; J.A. Armstrong; B.J. Marshall: "Campylobacter pyloridis gastritis and pectic ulceration." J. Clin. Pathol., 39: 353-365, 1986.

Johnston, B.; P. Reed; M. Ali: Prevalence of Campylobacter pylori in duodenal and gastric mucosa relationship to inflammation". Gut., 27:1132-1137, 1986.

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Ramírez Ramos, A.: Helicobacter pylori and gastroduodenal pathology. Lecture given at the XXII Pan American Congress of Digestive Diseases. Havana, Cuba, 1991.

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VERBRYCKE SYNDROME

Synonymy

S. of hepatic-gallbladder angle adhesion.

Syndromography

Clínica

This syndrome is characterized by pain in the right upper quadrant or epigastrium that the patient experiences when standing, accompanied by nausea.

Physical examination. Non-intense pain on palpation of the right quadrant, with muscular reaction (not constant).

Paraclinical examinations

Cholecystography. Normal function of the gallbladder. If a colon enema is performed simultaneously, the diagnosis can be specified.

Laparoscopy. It is the only method that allows direct visualization of the adhesions between the two organs, the reproduction of pain when the adhesions are touched or mobilized and in many cases also allows their section during the examination, thereby eliminating the clinical picture.

Syndromogenesis and etiology

Verbrycke syndrome is caused by adhesions between the gallbladder and the hepatic flexure of the colon.

Literature

Verbrycke, J.R., Jr.: “Adhesions of cholecystohepatic flexure, new syndrome with specific test.” JA.M.A., 114, 314-316, 1940.

WHIPPLE SYNDROME

Synonymy

S. of lipophagic intestinal granulomatosis. S. of intestinal lipodystrophy. S. of arthropericardial steatorrhea (see S. of malabsorption).

Syndromography

Clinic

Whipple syndrome predominates in men. It begins between the fourth and fifth decades of life with a picture of migratory arthralgias, diffuse abdominal discomfort, intermittent diarrhea with voluminous, foamy, foul-smelling stools, in addition, there is cough, dyspnea. Loss of strength and weight and fever intermittent.

Physical examination. Grayish pigmentation of the skin, occasionally purple. Edema. Emaciation. Abdomen: generalized discomfort; soft on palpation, sometimes ascites. In addition, the examination reveals lymphadenopathy and polyserositis.

Paraclinical examinations

Stool. Rich in fats and fatty acids.

Blood. Hypochromic anemia. Elevated erythrocyte sedimentation rate. Hypoproteinemia, hypocholesterolemia, normal induced hyperglycemia or a flat curve.

Gastroacidogram. Decreased free hydrochloric acid.

Jejunum biopsy. Club-shaped villi (one end thicker) and with a velvety appearance. Presence of macrophages at the level of the lamina propria, which contain PAS-positive material.

Node biopsy. Macrophages with the characteristics indicated above.

Radiology. Thickening of the folds in the jejunum and duodenum, with or without fragmentation or flocculation of the barium.

Polyserositis. Joint changes similar to rheumatoid arthritis.

Mediastinal lymphadenopathy. Abnormal abdominal lymphangiography with enlarged retroperitoneal nodes similar to defects found in lymphomas.

Syndromogenesis and etiology

This syndrome causes a malabsorption syndrome that explains most of the general and gastrointestinal disorders found.

The etiology is unknown.

Literature

Haubrich, W.S.; E.C. Raffensperger and R.W. Ammann,: Whipple's Disease. Bockus, H.L.: Gastroenterology. ed. 3. Philadelphia, Saunders, 1994.

Sleisenger, M.; J. Fordtran: Gastrointestinal diseases. t. II. 3rd ed, Pan-American Medical Ed. Mexico, 1985.

WILSON SYNDROME

Synonymy

S. de Kinnier-Wilson. S. of Westphal-Strumpell. S. of hepatolenticular

Syndromography

Clinic

Wilson syndrome predominates in men. Symptoms begin in childhood or young adulthood. The tremor begins in the limbs and spreads to the head and body.

It is accompanied by weakness, instability, dysarthria and mental disorders. In advanced states: anarthria and dysphagia.

Physical examination. In the eyes, a circular pigmentation is observed on the periphery of the cornea, at the level of Descemet's membrane, known as the Kayser-Fleischer ring, of a golden-greenish, sometimes reddish color that is characteristic of this condition, and which sometimes requires examination with the slit lamp to be able to observe it. There is also discoloration of the skin and sometimes jaundice, spider veins and hepatosplenomegaly, bone and joint alterations, muscle weakness, mild or severe incoordination, choreic movements, dystonic spasms, convulsions and plastic rigidity of the limbs.

Paraclinical examinations

Blood. Hypercupremia, hypoceruloplasminemia, below 20 mg/100 mL.

Liver function tests. Same as those of liver cirrhosis.

Urine. Hypercupruria greater than 50 w/24 h. Hyperminoaciduria.

Radiology. Osteoporosis.

Laparoscopy. A reduced liver size is observed with nodular alterations on the surface typical of cirrhosis that almost always take the form of medium-sized nodules.

Liver biopsy. Post-necrotic nodular cirrhosis, with active regeneration. Increased cellular copper content.

Syndromogenesis and etiology

It is a defect in copper metabolism that is deposited in the liver and gray nuclei at the base of the brain, which determines the extrapyramidal condition of these patients. The primary metabolic defect is an inability to maintain copper balance near zero. The capacity of hepatocytes to store copper can be exceeded, and the metal appears free in the blood and is taken up in extrahepatic locations.

The etiology is genetic due to a mutation of an autosomal recessive gene located at 13q14-q21, probably related to copper transfer enzymes as a hypothesis about the basic defect.

Literature

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Ouwen, R.H.J., et al.: "Close linkage of the Wilsons' disease locus to D13S12 in the chromosomal region 13q21 and not to ESD in 13q14." Hum. Genet., 85:560-562, 1990.

Westphal, C.: "Ueber eine dem Bilde der cerebro-spinalen graven Degeneartion ahnliche Erkrantomischen Befund, nebst einigen Beerkungen oe uber paradoxe Contraction." Arch. Psychiat. Berlin, 14:87-134, 1883.

Wilson, S.A.K.: "Progressive lenticular degeneration: a familial nervous disease associated with cirrhosis of the liver." Brain, 34:295-509, 1911-1912.

NON-GRANULOMATOUS CHRONIC ULCERATIVE JEYUNITIS SYNDROME

Synonymy

Chronic ulcerative idiopathic enteritis S. S. of jejunum-nonspecific chronic ulcerative ileitis.

Syndromography

Clinic

It presents with all the symptoms of a malabsorption syndrome in different degrees of progress (see malabsorption syndrome).

Paraclinical examinations

They are the same ones used in malabsorption syndrome, especially in non-tropical sprue. The jejunum biopsy is very valuable.

Syndromogenesis and etiology

It is known that it is different from non-tropical sprue, lymphoma, dysgammaglobulinemia, because it does not respond to a gluten-free diet, due to the type of pathological lesion (ulcerations of the jejunum with atrophy in the adjacent areas and normal mucosa); and on the other hand because there is a return to the normal protein level after treatment with corticosteroids. However, after several relapses he no longer responds to treatment.

The etiology is unknown.

Literature

Jeffries, GH.; H. Séinberg and M.H, Sleisenger: “Chronic ulcerative (nongranulomatous jejunitis)”. Am. J. Med., 44:47-59, 1968.

ZANCA SYNDROME

Syndromography

Clínica

This syndrome presents the symptoms of familial polyposis of the colon and is also associated with: multiple cartilaginous exostoses of the tubular bones and spine, sparing the ribs and skull.

Paraclinical examinations

bone radiology. Multiple osteochondromatous tumors with bone structure and cartilaginous coverage; While these lesions occur in early childhood, polyposis appears between the second and third decades.

The prognosis is guarded, since polyps in 70% of patients tend to become cancerous within 15 years.

Colonoscopy. It shows the polyps and is very necessary to try to eliminate them or monitor their evolution.

Syndromogenesis and etiology

It is a probably autosomal dominant inherited condition, although the exact mechanism of these alterations is unknown.

Bibliography

Zanca, P.: “Multiple hereditary cartilaginous exostoses with polyposis of the colon.” US Armed Forces Med. J. 116:116-120, 1956.

ZIEVE SYNDROME

Synonymy

S. of hyperlipemia-hemolytic anemia-jaundice. S. by Leslie-Zieve. S. pancreatic-hepatic.

Syndromography

Clinic

Zieve syndrome occurs in alcoholic patients and is characterized by mild jaundice accompanied by hemolytic type anemia and hyperlipidemia, with hypercholesterolemia.

The symptoms are: pain in the epigastrium or right hypochondrium, with frequent recurrences, anorexia, nausea, vomiting, weight loss. Fever and chronic diarrhea also occur. These symptoms are episodic and recurrent.

Physical examination. Hepatomegaly, arachnoid telangiectasias.

Paraclinical examinations

Blood. During crises, liver function tests (transaminases, phosphatase) are altered. Hyperbilirubinemia, hypercholesterolemia and hyperlipemia. Hyperuricemia, normal amylase. Hemolytic anemia, increased platelets, leukocytosis.

Bone marrow. Marked hyperplasia of normoblasts, presence of histiocytic phagocytes with lipid granules inside.

Liver biopsy. Mild to marked steatosis of the periportal connective tissue.

Syndromogenesis and etiology

Its exact mechanism is unknown but it is defined that alcohol is the cause of the syndrome, since this condition disappears if alcohol is stopped.

Literature

Schubotz, R.; K.M.Goebel und H. Kaffarnik: "Veränderungen in den Membranlipiden der Erythrocyten bei ähanolinduzierter Hyperlipidämie (Zieve-Syndrom)". Klin. Wschr., 54:827-833, 1976.

Zieve, L.: "Jaundice, hyperlipemia and hemolitic anemia: a heretofore unrecognized syndrome associated with alcoholic fatty liver and cirrhosis." Ann. Intern. Med., 48:471-496, 1958.

ZOLLINGER-ELLISON SYNDROME

Synonymy

Ström-Zollinger-Ellison S. Ulcerogenic S. due to pancreatic islet cell adenoma. S. of gastrinoma.

Syndromography

Clinic

This syndrome is a little more common in men than in women. It begins most of the time in the second to fifth decades; only 10% begin in the first two decades.

It is characterized by peptic ulcer with pain (90% of cases). The pain is intense and refractory to usual medical treatments and even surgical treatments with partial or subtotal resection of the stomach. There is hematemesis and/or melena (45%), vomiting (25%), intense diarrhea (36%), which can lead to dehydration and electrolyte alterations, steatorrhea, abdominal pain (50%). These symptoms are recurring. Pancreatic tumor rarely produces local symptoms.

Physical examination. Signs of dehydration in 60% of patients.

Paraclinical examinations

Gastroacidogram. Marked hypersecretion of chloric acid water.

Stool. Steatorrhea and diarrhea frequently occur, which disappear with sustained gastric aspiration.

Radiology. It shows multiple ulcers in abnormal locations (lower duodenum and jejunum). Marked hypertrophy of the gastric, duodenal and jejunal folds. Rapid evacuation of barium through the small intestine.

Ultrasound, computed tomography and selective arteriography. They only discover 20-309% of tumors, since these are generally small in size.

Determination of gastrin in blood. It is the most accurate test to ensure the diagnosis. In gastroduodenal ulcers, gastrin remains at a level of 40 to 50 pg/mL and does not usually exceed 150 pg/mL. In these cases it is always between 300-1,000 pg/mL and cases of 450,000 pg/mL have been reported. When there are doubts, an intravenous secretin test should be performed, which rapidly stimulates gastrin secretion.

Syndromogenesis and etiology

This syndrome is due to an adenoma of the islets of Langerhans originating in the delta cells, which produce gastrin, that is, a gastrinoma.

In 50% of cases the tumor is malignant and causes metastasis. Excessive production of gastrin is the cause of hyperacidity and increased gastric folds.

Literature

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Gerstein, J.D. and R.W. Muir: "Zollinger-Ellison syndrome. "A review". Amer. Surg., 41:230-239, 1975.

Zollinger, R.M. and E.H. Ellison: “Primary peptic ulcerations of the jejunum associated with islet cell tumors of the pancreas.” Ann. Surg., 142:709-728, 1955.

RELATED DIGESTIVE SYNDROMES

Cutaneous-intestinal syndromes

Amyloidosis.
Carcinoid.
Crohn's (with pyoderma gangrenosum).
Cronkhite-Canada.
Degos.
Dermatomyositis.
Scleroderma.
Gardner.
Grönblad-Strandberg-Touraine.
Blue ampullary nevus.
Peutz-Jeghers.
Rendu-Osler-Weber.

Syndromes with protein-losing enteropathy

Intestinal lymphangiectasia.
Malabsorption.
Menetrier.

Syndromes with exocrine pancreatic insufficiency

Clarke-Hadefield.
Comfort-Steinberg.
Fitz.
Kwashiorkor.
Mucoviscidosis.
Chronic pancreatitis.
Pancreatometaphyseal.
Schwachman.

Syndromes with sclerosing-lipogranulomatosis

Sclerosing cholangiitis.
Mesenteric panniculitis.

These syndromes represent the same pathological process, the difference in their clinical manifestations depends only on the location of the lesion.

Obstruction due to autogranulomatosis of the colon.
Ormond.

Syndromes with polyposis

Solitary adenoma.
Familial polyposis of the colon.
Colon polyposis with accessory symptoms:
a) Zanca (with cartilaginous exostoses).
b) Oldfield (with sebaceous cysts).
c) Turcot (with brain tumors).
d) Gardner (with skull osteoma, cutaneous fibromas, dermoid cysts), Werner.
e) Peutz-Jeghers.
f) Cronkhite-Canada.
g) Juvenile polyposis.
h) Bandler.
g) Bean.

Postgastrectomy syndromes

Aerophagia retching, belching.
Albatross
Iron deficiency anemia.
Megaloblastic anemia.
Blind handle.
Malnutrition.
Dumping.
Neurotic in gastrectomized patients.
Weight loss due to poor food intake.

Ischemic visceral syndromes

From the celiac trunk.
Of the superior mesenteric artery.
Of the median arcuate ligament.
Median aortic obliteration.
Ortner.
Mesenteric arteritis.
Abdominal migraine.
Visceral aorto-iliac steal.
Renal artery steal.

ABDERHALDEN-KAUFMANN-LIGNAC SYNDROME

Synonymy

S. of cystinosis. S. or cystine deposition disease. S. of Lignac. S. de Fanconi. S. of osteomalacia and rickets with glucoaminophosphaturic renal diabetes. S. de Lignac-Fanconi.

According to Bickel it is the same Fanconi syndrome (see this one), but DeToni considers two clinically separate conditions and recognizes two different syndromes:

1. Abderhalden-Kaufman-Lignac syndrome or cystine deposition disease.
2. DeToni-Debré-Fanconi syndrome or glucoaminophosphaturic renal diabetes with osteomalacia and rickets (see Fanconi-DeToni syndrome).

The most generalized criterion is that both syndromes are identical or very similar.

Syndromography

Clinic

The patient presents marked dwarfism, although proportionate (early symptom), especially of the metaphyses, with fractures and pseudofractures. Renal symptoms without increase in blood pressure or fundus. Thermal lability, with very high fever especially in the mornings (inverted febrile type). Adynamia in skeletal muscles (due to hypokalemia). Photophobia. Marked constipation (stipations are palpable in the colon).

Normal intelligence. Thymic hypoplasia.

Although the child is born apparently normal, in the following months his general condition deteriorates: anorexia, weight loss, vomiting, fever without apparent cause and a toxic state.

In the first years his condition worsens and symptoms of acute uremia and electrolyte disorders occur, leading to death almost always before the age of eight.

Some cases are less acute and progress to chronicity and adulthood.

Paraclinical examinations

Blood. Discreet anemia. In the more advanced periods: hyperchloremia, hypophosphatemia, decreased alkaline reserve, increased total proteins. Hypokalemia.

Medulogram. Typical birefringent tetragonal or hexagonal cysteine crystals are observed.

Urine. Alkaline, decreased density, aminoaciduria. Occasionally glucosuria with acetonuria. Cysteine crystals can be demonstrated with the Abderhalden II reaction.

Syndromogenesis and etiology

It is an autosomal recessive inherited condition. The causal mechanism has not been well clarified and the alterations

Metabolic disorders affect not only cysteine but other amino acids with the production of p-hydroxyphenylpyruvate and homogentisic acid.

Certain proteins that are not completely metabolized behave like a heterologous protein that damages the glomeruli and especially the glomerular membrane, producing glomerulonephrosis accompanied by tubular nephrosis, which causes nephropathy, arterial hypertension and uremia.

Cysteine deposits are constant in the bone marrow, spleen and lymph nodes and in general in the reticuloendothelial system.

Literature

DeToni, G: “Renal rickets with phosphoglucoamine renal diabetes (DeToni-Debré-Fanconi syndrome)”. Ann. Paediat., 187:42-80, 1956.

Fanconi, G.: “Die nicht diabeticischen Glycosurien und Hyperglykamien des lateren Kindes". Jahrb. F. Kinderh., 133:257-300, 1931.

Lignac, G.O.E.: Ueber Storung des Cystinstoffwechsel bei Kinder". Deutsch. Arch. f. klin. Med., 145:139-150, 1924.

ACHARD-THIERS SYNDROME

Synonymy

Bearded woman diabetes s.

Syndromography

Clínic

Voice change, oligomenorrhea or amenorrhea.

Physical examination. Facial hirsutism and acne, clitoral hypertrophy, breast atrophy, obesity, abdominal stretch marks, high blood pressure.

Paraclinical examinations

Blood. High blood glucose, decreased glucose tolerance

Nuida

Urine. Increase in urinary steroid excretion after ACTH stimulation.

Syndromogenesis and etiology

Hyperplasia or adenoma of the adrenal cortex with increased production of androgens and 1 1-oxysteroids.

Literature

Achard, C. and J. Thiers: Le virilisme pilaire et son association a l'insuffisance glycolytique (diabete des femmes a barbe)". Bull. Academic Nat. Med. Paris, 86:51-66, 1921.

XANTHURENIC ACIDURIA SYNDROME

See pyridoxine-dependent syndromes.

Syndromography

Clinic

It affects both sexes. Since childhood there is mental retardation, variable symptoms, moderate stomatitis or cheilosis, very frequent urticaria, anemia, bronchial asthma. These patients have diabetic family members.

Paraclinical examinations

Urine. Excessive excretion of xanthurenic acid, kynurenic acid, 3-hydroxykynurenine and kynurenine after a tryptophan load. Urinary abnormalities disappear temporarily with large doses of pyridoxine.

Syndromogenesis and etiology

This syndrome is due to a defect in chemoreninase, a vitamin B-dependent enzyme, in the tryptophan catabolic pathway.

Two forms are known: the one that responds to vitamin B, and the one that does not respond.

It is transmitted by autosomal recessive inheritance.

Bibliography

Knapp, A.: "Uber eine neue hereditare, von Vitamin-B6 abhangige Storung im Tryptophan-Stoffwechsel." J. Clin. Acta, 5:6-13, 1960

Tada, K.; Y. Yokoyama; H. H. Nakagawa, et al.: "Vitamin B, dependent xanthurenic aciduria." Tohoku J. Exp. Med., 93:1 15-124, 1967.

ACROMEGALIC SYNDROME

Synonymy

S. de Marie I. S. of anterior pituitary hypersecretion.

S. of eosinophilic adenoma.

Syndromography

Clinic

From a clinical point of view, two syndromes are distinguished:

1. Endocrine syndrome due to hormonal secretion.
2. Neurological syndrome due to compression mechanisms due to tumor growth.

Endocrine syndrome. Gradual and slow onset, generally between 30 and 50 years of age. It is characterized by: headache, pain in the dorsal region of the trunk, in the lower limbs, sweating, hypermenorrhea or amenorrhea, loss of libido and impotence in men. Sometimes there is a temporary increase in libido at the time of the onset of the disease, polyuria, polydipsia and muscle weakness.

Facial changes. Prognathism and separation of the teeth, thickening of the nose, prominence of the supraorbital edges and enlargement of the lips and tongue, thickening of the rough surfaces of the skull.

Changes of the trunk and lower limbs. Spade-shaped enlargement of the hands, fingers flattened at the tip and tassel-shaped hyperostosis in the terminal phalanges, changes in shoe size that impress the patient.

Dermal changes. Pigmentation, hirsutism, increased sebaceous secretion.

Kyphosis, muscle and joint hypertrophy. Decrease of the genitals. Tachycardia and hypertension.

Metabolic alterations. Hyperglycemia, glycosuria and increased basal metabolism.

Neurological syndrome. Low intensity headache, located bitemporally or retroocularly at first, due to increased pressure in the sella turcica, later the headache occurs due to increased intracranial pressure. Primary optic atrophy due to pressure on the optic nerves or the optic chiasm manifested by progressive pallor of the papilla, bitemporal hemianopia due to compression of the crossed nasal fibers of the chiasm, and in advanced cases complete blindness of one eye combined with temporal hemianopia of the other. When the tumor expands posteriorly it can cause homonymous hemianopia due to compression of an optic tract. In the case of very voluminous tumors, it can cause oculomotor paralysis and trigeminal neuralgia (ophthalmic branch) due to compression of these nerve trunks in the cavernous sinus. Suprasellar expansion of the tumor can cause symptoms of compression of the cerebral peduncles, diencephalon or temporal lobes, causing uncinate crises.

Paraclinical examinations

Blood. Diabetes mellitus. Increase in somatotropin, sometimes

Radiology. Increased bone silhouette of the skull, open sella turcica. The tomography may show a small tumor in the pituitary gland.

Campimetry.

Computerized axial tomography. Coronal sections of the sella turcica 1.5 mm thick allow adenomas to be visible and determine the extent of possible suprasellar extension, often revealing microadenomas.

Nuclear magnetic resonance. It is effective for visualization of adenomas.

Urine. Decreased FSH, increased 17-ketosteroids, glycosuria, creatinuria.

Syndromogenesis and etiology

It is due to an increase in the secretion of growth hormone after puberty that can be caused by two different forms: primary adenomatous pituitary acromegaly and hypothalamic acromegaly secondary to an increase in the production of growth-releasing factor (GRF) with secondary formation of an eosinophilic or intermediate-type adenoma.

A genetic etiology of autosomal dominant inheritance with reduced penetrance of the gene has been proposed.

Literature

Abbassioun, K., et al.: *Familial acromegaly wich pituitary adenoma: report of three affected sibling". J. Neurosurg., 64:510-512, 1986.

Marie, P. in Major, R.H.: Classic Descriptions of Diseases. ed. 2, Springfield, Thomas, 1939.

Paschkis, K.E.; A.E. Rakoff; A. Cantarow and J.J. Rupp: Clinical Endocrinology. ed. 3, New York, Harper & Row, 1967.

ADDISON SYNDROME - FAMILIAR JUVENILE HYPOPARATHYROIDISM IDIOPATHIC

Syndromography

Clínica

Endocrine system:

1. Hypoparathyroidism manifested by tettania.

2. Addison's disease (see this one).
3. Hypothyroidism.

Skin and annexes:

Dry skin and brittle hair. The eyebrows, pubic and axillary hair are noticeably diminished.

Nervous system:

Increased cerebrospinal fluid pressure, papilloedema, intracranial calcifications, epileptic seizures and mental retardation. Rigidity and paresthesias. Abdominal pains.

Oral manifestations:

Moniliasis that affects lips, tongue, oral mucosa, palate and larynx. The teeth are small and with pointed excavations.

Other findings:

Keratoconjunctivitis, photophobia, hoarseness, laryngospasm and wheezing.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

ADRENOGENITAL SYNDROMES

Synonymy

S. corticosexuals. S. adrenal virilizers. S. adrenal feminizers.

Syndromography

Clínic

Clinical findings depend on the etiology, which includes: congenital adrenal hyperplasia, feminizing adrenal tumors, virilizing adrenal tumors, and an ill-defined group with hirsutism with or without menstrual disorders (see these).

In the clinic, these syndromes form different production mechanisms independently of certain clinical peculiarities. We will analyze them below to be able to bring them together here.

CONGENITAL ADRENOGENITAL SYNDROME

Synonymy

S. de Crecchio. S. of congenital corticoadrenal hyperplasia.

Syndromography

It has four subtypes:

1. Simple virilizing.
2. Sodium loss. (See, in pediatric syndromes, the S. of congenital adrenal hyperplasia type II.)
3. Hypertensive.
4. 3-B-hydroxysteroid dehydrogenase defect. (See in pediatric syndromes the S. of congenital adrenal hyperplasia type II).

ADRENOGENITAL SYNDROME. FEBRILE FORM

Syndromography

Clínic

Recurrent fever in patients with adrenal hyperplasia syndrome.

Syndromogenesis and etiology

Excessive production of etiocholanolone.

ADRENOGENTTAL SYNDROME. HYPERTENSIVE FORM

Syndromography

Clínic

Moderate to severe hypertension in patients who present symptoms and signs of simple virilization. Some patients have only minor signs of virilization.

Paraclinical examinations

Biochemical study. Pregnantriol not very high.

Syndromogenesis and etiology

Congenital familial adrenal hyperplasia with severe 11-B-hydroxylase defect and excessive excretion of desOxicorticosterone and other cortisol precursors.

ADRENOGENITAL SYNDROME. SIMPLE VIRILIZING FORM IN FEMALES

Synonymy

S. de Apert-Gallais.

Syndromnography

Clinic

Rapid and progressive virilization from the moment of birth, clitoral hypertrophy that can lead to frequent erections. Deep voice, accelerated somatic growth.

Physical examination. The diagnosis is easier than in men since variable degrees of abnormalities of the external genitalia are present from birth, resulting in pseudohermaphroditism. The changes can range from minimal (simple growth of the clitoris) to maximum (when the girl is mistaken for a cryptorchid boy).

Armpit and pubic hair appear between 6 months and 3 years of age. Acne manifests early. Somatic growth accelerates. Solid muscles, thick hair on the beard and body, no breast development, amenorrhea.

Paraclinical examinations

Chromosomal and nuclear chromatin study. To define sex.

Examination of urethra and vagina.

Radiology and endoscopy.

Hormonal studies. See below on the male.

Syndromogenesis and etiology

Congenital family inheritance, autosomal recessive mutant gene, excess adrenal androgen secretion in fetal life.

ADRENOGENITAL SYNDROME. SIMPLE VIRILIZING FORM IN MEN

Syndromography

Clínica

Children of a few months with frequent erections, from 1-5 years the voice becomes deep, there is marked muscle development.

Physical examination. Early macrogenitosomia, within a few months enlargement of the penis with normal testicles, presence of axillary and pubic hair, increased skin pigmentation (like Addisonian), seborrhea, acne between 1 and 5 years, and between 8 and 10 years fusion of the epiphyses.

Paraclinical examinations

Urine. Increased 11-oxysteroids, 17-ketosteroids and pregnancyriol. Cortisone therapy decreases the size of the testicles and the excretion of 17-ketosteroids and pregnancyriol (the latter makes the differential diagnosis with virilizing tumors). The administration of ACTH markedly increases the excretion of pregnancyriol and steroids already mentioned, and moderately increases androgens and estrogens.

Cortisol metabolites are absent or there is insufficient response to ACTH (pathognomonic). Administration of mepirapone increases the excretion of pregnantriol and 17- -ketosteroids.

Syndromogenesis and etiology

See this syndrome in females.

NON-TUMORAL ADRENOGENITAL SYNDROME WITH HIRSUTISM IN FEMALES

Syndromography

Clinic

Non-tumor adrenogenital syndrome with hirsutism predominates in females. Generally, puberty is normal, menstruations can be normal or abnormal. Some patients become pregnant and their offspring are normal. There is accelerated growth in childhood with premature closure of the epiphyses that results in short stature in adults.

Physical examination. It is important to know the date of appearance of the signs. Hirsutism of varying degrees is seen, isolated or associated with various forms of masculinization and acne.

Paraclinical examinations

Urine. There may be an increase in pregnancyriol, a moderate increase in 17-ketosteroids. ACTH stimulation: increase in the excretion of pregnancyriol and corticosteroids. If cortisone is administered, there is a decrease in pregnancyriol.

A subgroup can be recognized if increased urinary excretion of DHEA and other androgenic steroids is found.
This increase in DHEA excretion with hirsutism and menstrual disorders can be found in:

1. Moderate form of congenital adrenal hyperplasia.
2. Simple obesity.
3. Cushing syndrome.
4. Acromegaly and mental disorders.

Syndromogenesis and etiology

In some cases, partial 21-hydroxylase deficiency and/or 11-hydroxylase defect is suggested; In both forms, demonstrated or not, hormonal abnormalities are often familial.

It is a syndrome of genetic etiology and autosomal recessive inheritance.

NON-TUMORAL ADRENOGENITAL SYNDROME WITH HIRSUTISM IN MEN

Syndromography

Clinic

Asthenia, sterility.

Paraclinical examinations

Laboratory exams. Low sperm count.

Urine. Increased 17-ketosteroids and sometimes estrogens.

Testicular biopsy.

Syndromogenesis and etiology

Moderate adrenal hyperplasia. There may be a partial 21-hydroxylase defect.

Literature

Decurt, J.; M.E Jayle and P. Mauvais-Jarvis: "Virilisme surrenalien para ou post-pubertaire avec pregnanetriolurie." Rev. Eur. Endocr. 1:17, 1964.

Jayle, M.E.; D. Malassis and H. Pinaud: "Excretion de la dehydroepiandrosterone avant et apres administration d'ACTH en pathologie surrenalienne." Acta Endocr. (Kobenhavn), 31:1-32, 1959.

ADRENOGENITAL SYNDROME. FEMINIZING ADRENAL TUMORS

Syndromography

Clinic

Child (5-7 years old): gynecomastia, accelerated development of bones and muscles.

Girl (3-4 years): precocious puberty, breasts, axillary and pubic hair, vaginal bleeding, accelerated bone and muscle development.

Male adult (25-66 years): decreased libido and potency, pain at the tumor site, gynecomastia with or without sensitivity, testicular atrophy, areolar pigmentation and rarely discharge, prostate and penis generally normal and rarely atrophic, feminine hair changes.

Paraclinical examinations

Computerized axial tomography.

Urine. Estrogens are usually very high, 17-ketosteroids increased in most cases, pregnancyriol and pregnadiol almost always very high, 17-hydroxycorticosteroids increased in half of the patients.

ACTH stimulation. Negative, Cortisone suppression. Negative.

Syndromogenesis and etiology

Carcinoma or adrenal adenoma.

ADRENOGENITAL SYNDROME. VIRILIZING ADRENAL TUMORS

Syndromography

Clinic

Prepubertal female (from a few months until puberty): hirsutism, clitoral hypertrophy, rapid development of bones and muscles, no breast development, short stature, no menstruation at pubertal age.

Prepubertal male: early macrogenitosomia (for other manifestations see congenital adrenal hypertrophy).

Adult female (generally, between 30 and 40 years or later, even postmenopausal): hirsutism, beard, mustache and hair loss, acne, voice changes, oligomenorrhea progressing to amenorrhea, clitoral hypertrophy, muscle hypertrophy, loss of fat deposits, breast atrophy, increased libido.

Paraclinical examinations

Computed axial tomography. It can show the tumor.

Urine. increased 17-ketosteroids (with DHEA more than half of the 17-ketosteroids).

ACTH stimulation. Negative.

Cortisone suppression. Negative.

Syndromogenesis and etiology

Carcinomas and adenomas in the adrenal gland.

Literature

Baulieu, E.E.; E Peillon and CJ. Migeon: Adrenogenital syndrome. In Eisenstein, A.B.: The Adrenal Cortex. Boston, Little- -Brown. 1967.

HARD WATER SYNDROME

Synonymy

S. of hypercalcemia during extracorporeal dialysis.

Syndromography

Clínic

This syndrome occurs during or after hemodialysis. It is characterized by nausea, vomiting, hot skin, extreme weakness, lethargy, blood pressure changes.

Occasional coagulation within the arteriovenous cannula.

Paraclinical examinations

Blood. High calcium and magnesium.

Analysis of the water used in dialysis.

Syndromogenesis and etiology

It occurs when the water softening process is abruptly discontinued while hemodialysis is being performed.

Literature

Freeman, R.M.: RL. Lawton and M.A. Chamberlain: “Hard water syndrome.” New Eng. J. Med., 276:113-118, 1967.

Schulten, H.K.; H.G. Sieberth; K.A. Deck, et al: "Das akute Hypercalcamiesyndrom als Dialysezwischenfall." Deutsch. Med. Wschr., 93:387-390, 1968.

AHUMADA-DEL CASTILLO SYNDROME

Synonymy

S. de Argonz-del Castillo. S. of amenorrhea-galactorrhea- -decrease in FSH.

Syndromography

Clinic

Unrelated to pregnancy (the opposite of Chiari-Frommel syndrome). Presents galactorrhea, amenorrhea and headache.

Physical examination. Normal breasts (in size, areola and nipple), galactorrhea, normal sexual characteristics.

Paraclinical examinations

Urine. Very low or absent urinary gonadotropin.

Radiology. Normal Turkish chair.

Syndromogenesis and etiology

Hypothalamic-pituitary functional alteration.

Literature

Ahumada, J.C. and E.B. del Castillo: "On a case of galactorrhea and amenorrhea." Bol. Soc. Obst. Gin., Buenos Aires, 11:64-72, 1932, 1:64-72, 1932.

Argonz, J. and E.B. del Castillo: "A syndrome characterized by estrogenic insufficiency galactorrhea and decreased urinary gonadotropin." J. Clin. Endocrinol., 13:79-87, 1953.

HEREDITARY ALACTASIA SYNDROME

Synonymy

Alactasis syndrome (see sugar-cleaving enzyme deficiency syndromes)

Syndromography

Clinic

It affects both sexes. It appears after the first feeding of milk. There is severe diarrhea, insufficient progress.

Paraclinical examinations

Stool. Lactose, organic acids, mainly acid lactic.

Syndromogenesis and etiology

Congenital lactase deficiency, autosomal inheritance recessive.

Literature

Holzel, A.: V. Schwarz and K.W. Sutcliffe: "Defective lactose absorption causing malnutrition in infancy." Lancet, 1:1126- -1128, 1959.

Milue, M.D.: "Hereditary abnormalities of intestinal absorption." Br. Med. Bull., 23:279-284, 1968.

ALBRIGHT SYNDROME I

Synonymy

S. McCune-Albright. S. of fibrous osteodystrophy. S. of disseminated osteitis fibrosa. S. with brown spots. S. de Fullen- - Albright. S. de Albright-Mc Cune-Sternberg. S. of bone fibrous dysplasia.

Syndromography

Clinic

It occurs in children or young adults, it predominates in the female sex.

The symptoms are very variable. Skeletal injuries are usually discovered due to deformity or fractures. Symptoms attributable to bone disease are: headache, seizures, cranial nerve abnormalities, deafness, narrowing of the external auditory canal, or even scalp hemorrhages if there is craniofacial bone disease.

There is sexual precocity in some women and even less frequently in men; it is the presenting symptom before bone lesions. The main signs are: premature vaginal bleeding, breast development and the appearance of axillary and pubic hair.

In addition, there are "café au lait" spots on only one side of the body, with irregular (toothed) edges, unlike the smooth edge in neurofibromatosis; they almost never exceed six and the size varies from 1 cm to those that cover very large areas, especially on the back, buttocks and sacral region. When the lesions are found on the scalp, the hair at the level of the lesions is darker.

Cutaneous osteomas are accompanied by localized alopecia and there is a certain tendency to correspond with skeletal lesions; the pigmentation may be on the same side as the skeletal lesions and in fact cover them.

There is difficulty walking, pain in the legs and pathological fractures.

Other associated abnormalities: hyperthyroidism, less common, Cushing's syndrome, perhaps hypogonadotropic hypogonadism, and soft tissue myxomas. Facies leonti-
na. Short stature.

Physical examination. Bone deformities and pathological fractures, especially in the lower limbs and hip. Brownish pigmentation of the skin, of variable size with predominance in the head, neck, sacral and gluteal region. In the monostotic variety there is unilateral exophthalmia, unilateral optic atrophy, hearing loss, seizures, mental retardation.

Paraclinical examinations

Blood. Elevated phosphatase, normal calcium and phosphorus.

Urine. Normal excretion of 17-ketosteroids and FSH.

Radiology. Radiolucent area with a well-defined smooth or scalloped edge accompanied by focal thinning of the cortex.

The bone may be larger than normal. The lesions are not cystic, sometimes they appear multiloculated. Image on frosted glass. Coxa vara, shepherd's crook femur, bowing of the tibia, protrusion of the acetabulum.

Syndromogenesis and etiology

The cause is unknown and although the disorder is sporadic, family trees compatible with autosomal dominant inheritance have been observed. Cushing's syndrome can depend on abnormal ACTH production or adrenal adenomas. The bone lesion resembles that of hyperparathyroidism, and histological, but not clinical, parathyroid hyperplasia has been described.

G proteins are made up of three subunits, B and T, and couple hormone-receptor complexes with an intracellular effector; In the case that the effector is adenyl cyclase, there are two types of alpha extremities: the inhibitory one (alpha i) and the stimulatory one (alpha s). In Albright I syndrome, the mutation causes activation of the stimulatory G alpha protein.

Historically Fuller Albright described two defects of the same gene: mutation due to deficiency in the case of pseudohypoparathyroidism and mutation due to excess in the case of the syndrome in

The gene has been assigned to chromosome 20q12-q13.2. and autosomal dominant inheritance.

Literature

Albright, F.: A.M. Bufler; A.0. Hampton, et al.: "Syndrome characterized by osteitis fibrosa disseminata, areas of pigmentation and endocrine dysfunction with precocious puberty in females." N. Engl. J. Med., 216:727-746, 1937.

Gopal Rao, V.V.N.; S. Schnittger and I. Hansmann: “G protein Gs- alfa (G NAS1), the probable candidate gene for Albright hereditary osteodystrophy, is assigned to human chromosome 20g 12-q13.2". Genomics 10:257-261, 1991.

McCune, D.J. and H. Bruch: Osteodystrophia fibrosa". Amer. J. Dis. Child., 54:806-848, 1937.

METABOLIC ALKALOSIS SYNDROME

Syndromography

Clinic

It is characterized by tetany crisis that occurs only when serum calcium decreases with a normal or high pH and/or hyperkalemia. If there is hypokalemia, muscle weakness and paralytic ileus occur (see hypokalemia syndrome).

Paraclinical examinations

Blood. High pH, pCO, normal, low chlorine, high bicarbonate, normal or high sodium, low or normal potassium.

Urine. Alkaline, if there is loss of potassium it is acidic.

Syndromogenesis and etiology

The syndrome is caused by:

1. Excessive database administration.
2. Loss of chlorides (vomiting, diarrhea, diuretics).
3. Excessive loss of potassium.
4. Consequence of long-treated hypercalcemia (see this one).

Literature

Goldberger, E.: A Primer of Water, Electrolyte and Acid-Base Syndromes. ed. 3, Philadelphia, Lea && Febiger, 1965.

ALLAN-DENT SYNDROME

Synonymy

S. of arginosuccinuria.

Syndromography

Clinic

Normal patient at birth. History of inconstant vomiting in the first weeks of life. Normal acquisition of early skills, except language development. Between 1 and 2 years of age, mental retardation becomes evident and seizures and ataxia appear.

Physical examination. Tufted and brittle hair.

Hepatomegaly.

Paraclinical examinations

Blood. Arginosuccinase deficiency; normal arginosuccinic acid and urea.

Urine. Increased elimination of arginosuccinic acid.

Cerebrospinal fluid. Increased concentration of arginosuccinic acid.

Electrocardiogram.

Syndromogenesis and etiology

Genetic arginosuccinase deficiency.

Three clinical types are recognized: neonatal, infantile and chronic, possibly due to allelic mutations. It presents autosomal recessive inheritance.

Literature

Allan, JD., et al.: *A disease, probably hereditary, characterized by severe mental deficiency and a constant gross abnormality, of amino acid metabolism." Lancet, 1:182-187, 1958.

Efrom, M.L.: Diseases of the urea cycle. In Stanbury, J.B., J.B. Wyngaarden, and D.S. Frederickson: The Metabolic Basis of Inherited Disease, New York, Mc Graw-Hill, 1966.

AMYLOIDOSIS SYNDROME

Syndromography

Clinic

The symptoms are related to the organs involved, which is why it can begin as: nephrotic syndrome, renal failure, heart failure, arrhythmias, chronic malabsorption syndrome, intestinal obstruction, carpal tunnel syndrome, peripheral neuropathy, endocrine hypofunction, respiratory failure, hepatosplenomegaly, arthropathies, ecchymosis.

Paraclinical examinations

Congo red test. Positive.

Fine needle biopsy of abdominal fat.

Gingival, renal, rectal biopsies.

Urine. Proteinuria.

Protein electrophoresis. Inversion of the serine-globulin index, elevation of alpha 2.

Blood. Anemia, elevated or normal erythrocyte sedimentation rate.

Syndromogenesis and etiology

It is due to infiltration of tissues by an insoluble protein or protein complex. In the primary form and in the form associated with plasmacytosis it is a light chain immunoglobulin. In the secondary and hereditary form, the protein is not an immunoglobulin.

We will describe in the table the most accepted forms of amyloidosis according to the modern classification.

SHAPE	CLINICAL TYPE	DEPOSIT SITE
Familiar	Amyloid polyneuropathy (Portuguese, dominant inheritance)	Peripheral nerve, viscera
	Familial Mediterranean fever (recessive character)	Liver, spleen, adrenal
Generalized	Primary	Tongue, heart, intestine, muscles: smooth and striated, nerves, skin,
	Associated with plasma cell dyscrasia	Liver, spleen, kidneys, heart, adrenals
	Secondary: infection inflammation, rheumatoid arthritis, Hodgkin's, Crohn's, hypernephroma, leprosy, excessive use of intravenous drugs	Fur
Located	lichenified	Fur
	Endocrine diseases: thyroid carcinoma, diabetes mellitus, Addison	endocrine organ
Senile	Located	Heart, brain, pancreas

AMYLODOSE SYNDROME

HEREDOFAMILIAR

Syndromography

Clinic

Some authors highlight the importance of predominantly one organ lesion to identify the entity as neuropathic, nephropathic or cardiopathic amyloidosis. Others highlight genetic aspects.

Neuropathic:

Type I. Lower limb neuropathy (due to familial amyloid).

It is accompanied by progressively severe neuropathy.

Patients have been found with scalloped or irregular pupils, bilaterally.

Type II. Found in families of Swiss and German origin.

It is less serious and is accompanied by carpal tunnel syndrome and vitreous opacities.

Type II. Severe, generalized variety accompanied by renal amyloidosis.

Type IV. With cranial nerve neuropathy and corneal web dystrophy. Hereditary cerebral amyloidosis with hemorrhage.

Nephropathic:

Familial Mediterranean fever with fever, abdominal pain. Chest or joint pain that precedes or accompanies renal amyloidosis.

Amyloidosis is the first or only manifestation.

Cardiopathic:

Severe familial amyloid heart disease.

Paraclinical examinations

The specific diagnosis is established through a tissue sample obtained by biopsy and the appropriate stains (Congo red) and study with a polarized microscope.

Syndromogenesis and etiology

Heredofamilial amyloidosis has an autosomal dominant hereditary mechanism, except in familial Mediterranean fever in which there is an autosomal recessive disorder and the amyloid is type AA.

The affected organs are infiltrated by an eosinophilic, hyaline amorphous substance (amyloid).

Literature

Harrison: “Principles of Internal Medicine.” Inter-American Ed., MeGraw Hill (eleventh edition), 1988.

Kyle, R.A. "Amyloidosis (Al). Clinical and Laboratory Features in 229 cases". Mayo Clinic Proc., 58: 665, 1983.

ANALBUMINEMIC SYNDROMES

Three congenital alterations of serum albumin are recognized:

1. Analbuminemia.
2. Bisalbuminemia.
3. Abnormal albuminemia.

ANALBUMINEMIA SYNDROME

Syndromography

Clinic

Moderate edema.

Paraclinical examinations

Blood. Low serine, accelerated erythrocyte sedimentation rate, high cholesterol, low calcium.

Liver function tests. Normal.

Syndromogenesis and etiology

Analbuminemia is a rare disorder of autosomal recessive inheritance, in which homozygous individuals show an absence of serum albumin.

There is great genetic heterogeneity, generated fundamentally by base changes and amino acid substitution.

Literature

Takahashi, N. et al. “Aminoacid substitutions inherited albumin variants from Amerinchan and Japanese populations.” Proc. Nat. Acad. Sci., 84:8001-8005, 1987.

BISALBUMINEMIA SYNDROME

Syndromography

Clinic

Asymptomatic.

Paraclinical examinations

Blood. Protein electrophoresis shows normal albumin and an additional abnormal fraction.

Syndromogenesis and etiology

Although it is unknown, it has characteristics of autosomal dominant inheritance with completely expressed normal and abnormal genes.

ABNORMAL ALBUMINEMIA SYNDROME

Possibly non-specific congenital abnormality associated with goiter and deafness syndrome that is characterized by abnormal albumin in the blood.

Literature

Bennhold, H.: H. Peters and E. Roth: “Uber einen Fall bon kompletter Analbuminaemie ohne wesentliche klinische Krankheitszeichen.” Verh. Deutsch. Ges. Inn. Med., Kong, 60:630-634, 1954.

Scheurlen, P.G.: "Uber Serumeiwessveranderungen beim Diabetes mellitus." Klin. Wcschr., 33:198-205, 1955.

ANOVULATION SYNDROME AFTER THE USE OF ORAL CONTRACEPTIVES

Synonymy

S. of oversuppression.

Syndromography

Clinic

Amenorrhea of 3 months' duration, or marked irregularity of menstrual cycles, infertility after discontinuation of oral contraceptive therapy.

Paraclinical examinations

Determination of FSH, LH and estrogen level.

Syndromogenesis and etiology

Probable dysfunction of the hypothalamic centers in relation to the release of gonadotropins.

Literature

Horowitz, B.J.; M. Solomkin and S.W. Edelstein: "The oversuppression syndrome." Obstet. Gynec., 31:387-389, 1968.
Whitelow, M.J.; V.. Nola and C.E Kalman: "Irregular menses, amenorrhea, and infertility following synthetic progestational agents." J.A.M.A., 195:780-782, 1966.

ACUTE GOUTY ARTHRITIS SYNDROME

Syndromography

Clínica

It appears more frequently in men between the third and fourth decades of life; in women after menopause.
It frequently begins during the night, with severe, deep pain, like a bite, that is located in the thick joint, less frequent in other places: ankle, heel, knee, wrist.

Great discomfort, chills, tremors; there is no improvement with position changes.

Physical examination. Inflammatory signs in the affected area.

Paraclinical examinations

Blood. Hyperuricemia.

Syndromogenesis and etiology

It is due to a disorder of purine metabolism. Primary gout has an autosomal dominant or sex-linked familial character. Secondary gout appears in multiple diseases: hematological (myeloproliferative syndromes, hemolytic anemia), iatrogenic (drugs that increase uric acid), obesity, wasting, chronic renal failure, glycogen storage disease (type I), sarcoidosis, psoriasis.

Literature

Galen (131-200 A.D.) quoted by Newirth, E.: "Milestones in the diagnosis and treatment of gout." Arch. Int. Med., 72:377-387, 1943.

Sydenham, T.: The works of Thomas Sydenham, trans. by R.G. Latham. V.I, p. 214. Sydenham Society, London, 1850.
Wyngaarden, J.B.: Gout. In: Stanbury, J.B., J.B. Wyngaarden, and D.S. Frederickson: The Metabolic Basis of Inherited Disease.
ed. 2. New York, McGraw-Hill., 1966.

CHRONIC GOUTY ARTHRITIS SYNDROME

Syndromography

Clínica

For the syndrome to establish itself, it takes an average of 10 or 12 years after the first acute attack. The beginning zo can be insidious or a sequel to repeated attacks. It is common that when the chronic form develops, acute attacks disappear.

Physical examination. Polyarthritis with eventual evolution to grotesque, destructive, ulcerated forms. Tophi in the ear cartilage, tendons, and less frequently skin, nose and eyelids. With ulceration of the tophus, a material reminiscent of clay (a material similar to clay or sand) is excreted and the pain is relieved.

Paraclinical examinations

Blood. Hyperuricemia.
Radiology. Joint deformity, osteolysis areas.
Joint puncture. Synovial fluid, presence of uric acid crystals.

Syndromogenesis and etiology

See acute form.

There may be kidney, heart or brain complications that depend on the deposition of urate in these tissues.

Literature

Galen (131-200 A.D.) quoted by Newirth, E.: "Milestones in the diagnosis and treatment of gout." Arch. Int. Med., 72:377- -387, 1943.

Sydenham, T.: The works of Thomas Sydenham, trans. by R.G. Latham. V. II, p. 214. Sydenham Society, London, 1850.
Wyngaarden, J.B.: Gout. In: Stanbury, J.B., J.B. Wyngaarden and D.S. Frederickson: The Metabolic Basis of Inherited Disease.
ed. 2, New York, McGraw-Hill, 1966.

UNILATERAL GENITAL ATRESIA SYNDROME
AND DYSMENORRHEA

Syndromography

Clinic

It is observed in patients whose ages range between 12 and 27 years. Dysmenorrhea appears shortly after menarche and increases in severity with subsequent menstruations.

Other accompanying symptoms may be: nausea, vomiting, constipation or diarrhea, dysuria.

Physical examination. Unilateral pelvic tumor.

Paraclinical examinations

Ultrasound.

Syndromogenesis and etiology

Incomplete fusion or poor adaptation of the Müllerian ducts causes unilateral atresia with accumulation of menstrual flow on that side with each menstruation, producing a cystic mass.

Literature

Merckel, G.C.; MC Sucoff and B. Sender: "The syndrome of dysmenorrhea and unilateral gynatresia in a double uterus: report of a case and review of literature." Amer. J. Obstet. Gynecol., 80:70-75, 1960.

Robinson, E.M.: *Supernumerary uterus in a girl eighteen years old: removal; recovery". Alabama Med. J., 23:152-154, 1910-1911.

BIEMOND SYNDROME I

Synonymy

Hypothalamic Biemond's S.

Syndromography

Clinic

Homogeneously distributed obesity, secondary hypogonadism, with impotence. Postaxial polydactyly especially in the feet and bilateral coloboma of the iris. Intermittent oliguria.
Attacks of vertigo and lipothymia. Psychic and oligo-alterations
phrenia

Syndromogenesis and etiology

There are few families reported. The presence of coloboma of the iris in four generations of a family and of postaxial polydactyly of the feet in the father and paternal uncles of cases with all the defects of the syndrome suggest an autosomal dominant mutation with variable expressivity.

The etiology is genetic.

Literature

Biemond, A.: "Het syndrom van Laurence-Biedl in een aanverwant, nieuw syndroom." Ned. Tschr. Geneesk., 78:1801- - 1814. 1934.

BIGLIERI SYNDROME

Synonymy

S. of 17-hydroxylase deficiency.

Syndromography

Clinic

This is an extremely rare condition, very few cases have been reported in the international literature, all in females. Primary amenorrhea, delayed or absent development of secondary sexual characteristics, fatigue, marked episodic muscle weakness, numbness and tingling in the limbs, and partial hair loss are observed. There is no development of the breasts, little body hair, infantile uterus, small ovaries to palpation, facies with wrinkles and signs of aging, all accompanied by hypertension. The response to treatment is optimal.

Paraclinical examinations

Blood. Hypokalemia, alkalosis. Low levels of cortisol, testosterone and androsterone. ACTH markedly increased.

Urine. Increase in I1-oxycorticosteroids, 17-ketosteroids apparently normal, pregnancyriol is markedly decreased. Low excretion of estrona and estradiol, increase in urinary gonadotropin, increase in the production of deoxycorticosterone, corticosterone, progesterone and
aldosterone.

Electrocardiogram. Signs of hypokalemia.

Syndromogenesis and etiology

It is a congenital defect in the activity of 17-hydroxylase, which causes a reduction in the synthesis of cortisol, estrogens and androgens.

Literature

Biglieri, E.G.; M.A. Herron and N. Brust: "17-hydroxylation deficiency in man." J. Clin. Invest.. 45:1946-1954, 1966.

Goldsmith, O., D.H. Solomon and R. Horton: "Hypogonadism and mineralocorticoid excess. The 17-Hydroxylase deficiency syndrome." New English J. Med., 277:673-677, 1967.

BRISSAUD I SYNDROME

Synonymy

S. de Brissaud-Meige. Brissaud's infantilism.

Syndromography

Clinic

Little development of the skeletal system, round face, prominent abdomen, thickened panicle, little genital development, absence of permanent teeth, absence of body hair.

Syndromogenesis and etiology

It constitutes a type of pituitary dwarfism.

Literature

Brissaud, P.: "L'infantilisme vrai." N. Inconog de la Salpetriere, Paris, 20:1,1907.

BÜRGER-GRÜTZ SYNDROME

Synonymy

S. of familial hyperlipoproteinemia type I. S. of familial hyperchylomicronemia. S. of familial fat-induced hyperlipidemia. S. of idiopathic hyperlipidemia. S. of hyperlipidemia with lipoid hepatosplenomegaly. S. of essential familial hyperlipidemia. S. of acute abdominal hyperlipidemia.

Syndromography

Clinic

It affects both sexes, of early onset, and becomes evident as soon as the child begins to ingest fat. There are attacks of abdominal pain, discomfort and anorexia, without nausea or vomiting; attacks may be accompanied by fever.

Physical examination. Eruptive xanthomas anywhere, including the mucous membranes, consisting of yellowish nodules with an erythematous base that fade and disappear in a few weeks. Signs of peritoneal irritation. Hepatosplenomegaly, sensitive splenic area. The crises are triggered by the ingestion of fats.

Paraclinical examinations

Blood. Hyperlipidemia following fat ingestion;
leukocytosis during crises. Normal liver function.

Syndromogenesis and etiology

It is a syndrome of genetic etiology with autosomal dominant inheritance.

The basic defect consists of deficiency of the enzyme lipoprotein lipase.

Literature

Bürger, M. and O. Grutz: “Ueber hepatosplenomegale lipoidose mit xanthomatsen veranderungen in haut und schleimhaut.”
Arch. Derm. Syph., 166:542-575, 1932.
Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The Metabolic Basis of Inherited Disease. New York, McGraw-Hill, 1966.

CHIARI-FROMMEL SYNDROME

Synonymy

S. de Frommel-Chiari.

Syndromography

Clinic

Persistent galactorrhea and amenorrhea several months after discontinuing breastfeeding. Headache, back pain, emotional instability, abdominal pain, small uterus.

Rarely decreased visual field.

Paraclinical examinations

Blood. Increased luteotropin.

Radiology. Enlarged Turkish chair.

Syndromogenesis and etiology

This is hypogonadism secondary to dysfunction, or more rarely to a pituitary tumor.

Literature

Chiari, J.B.; C. Braum and J. Spaetch: Report of diseases of women observed during the years, 1848 to 1855, inclusive, in the Department of Gynecology (Municipal Clinic). Vienna Klin. Geburtsh. V. Gynak. p.371, 1855.

Frommel, R.: "Ueber puerperale Atrophie des Uterus". Ztschr. Geburtsh. u. Gynak., 7:305-313, 1882.

CYSTATHIONURIA SYNDROME

Syndromography

Clinic

It is a rare condition, which presents mental retardation, developmental defects (small ears, clubfoot), pituitary diseases and in some cases acromegaly. Seizures, thrombocytopenia, and kidney stones may also occur.

Paraclinical examinations

Blood. Marked increase in cystathionine that is even detected in the cerebrospinal fluid.

Urine. Elimination of cystathionine.

Renal ultrasound and descending urogram.

Syndromogenesis and etiology

There is probably a deficiency of the enzyme cystathioninase that divides cystathionine into cysteine and homoserine.

Some cases with elevated cystathionine levels have been asymptomatic, so the relationship between the metabolic defect and the malformations is not clear.

The cystathioninase gene has been located on chromosome 16.

The basic defect is of genetic etiology and inheritance autosomal recessive.

Literature

Harris, H.; L.S. Penrose and D.H.H. Thomas: Cystathioninuria."
Ann, Human Genet., 23:442, 1959.
Frimpter, G.W.: Cystathionuria. In J.B. Stanbury; J.B. Wyngaarden, and D.S. Frederickson: The Metabolic Basis of Inherited Disease.
New York, McGraw-Hill, 1966.

CYSTINURIA SYNDROME

Syndromography

Clínica

The male is affected more severely than the female. Renal lithiasis around 30 years of age in half of the cases. Most patients are short and have loss of at least one essential amino acid, lysine.

Paraclinical examinations

Urine. Excessive excretion of amino acids (cystine, lysine, arginine and ornithine), cystine stones, hematuria, proteinuria.

Ultrasound and urogram.

Syndromogenesis and etiology

It is a defect of genetic etiology with allelic and non-allelic heterogeneity. It is estimated that some types are the result of compound genotypes, that is, heterozygous for two abnormal genes.

Three genetic types of cystinuria are described:

Type I, in which the homozygous individual excretes large amounts of cystine, lysine, arginine and ornithine in the urine, and the heterozygous parents do not present aminoaciduria.

Type I, in which heterozygous individuals may have cystine stones and a moderate degree of aminoaciduria with excretion of cystine and lysine.

Type II, in which there is a deficiency of intestinal transport of dibasic amino acids in the heterozygous individual.

Mutant genes of types I and II are allelic forms.

This syndrome should not be confused with cystine deposition syndrome (see this one) and other syndromes where cystinuria is part of a general aminoaciduria (De Toni-Fanconi syndrome, Wilson syndrome).

Literature

Garrod, A.E.: The croonian lectures on inborn errors of metabolism.
Lancet, 2:1908 (Lecture I. p. 1; Lecture II, p. 73; Lecture II, p. 142; Lecture IV, p. 214).
Knox, W.E.: Cystinuria. In Stanbury, J.B., J.B. Wyngaarden, and D.S Frederickson,: The Metabolic Basis of Inherited Disease.
New York, McGraw-Hill, 1966.
Stephens, A.D.: "Cystinuria and its treatment: 25 years experience at St. Bartholomew's Hospital." J. Inherit. Metab. Dis., 12:197-209, 1989.

CITRULINEMIA SYNDROME

Syndromography

Clinic

The syndrome manifests itself at an early age. Physical and mental development is normal until the disease becomes apparent.

manifest. Vomiting, irritability, seizures, difficulty progressing, mental retardation.

Paraclinical examinations

Blood. Postprandial hyperammonemia, normal urea; elevated citrulline in blood, urine and cerebrospinal fluid.

Electrocardiogram.

Syndromogenesis and etiology

It is due to a biochemical defect.

Literature

McMurray, W.C.. JC. Rathbun: Mohyuddin, et al.: "Citrullinuria." Pediatrics, 32:347-357, 1963.

Morrow, G.; THE. Barness and M.L. Efron: "Citrullinemia with defective urea production." Pediatrics, 40:565-574, 1967.

MALE CLIMATERIAL SYNDROME

Synonymy

Andropause.

Syndromography

Clínica

It generally occurs in many men after the age of 50-60, or later, and is not as frequent as the female climacteric. It is characterized by: depression, insomnia, fatigue, decrease and slowing of mental activity.

Anxiety, fear, apprehension, trembling. Vasomotor symptoms, alternating appearance of suffocation and sweating. Decreased libido and sexual potency.

Paraclinical examinations

Urine. 17-ketosteroids and gonadotropins usually normal; In rare cases its positivity confirms the climacteric condition.

Syndromogenesis and etiology

Opinions about the existence of this entity are contradictory. It is generally believed to be due to emotional disorders rather than true hormonal problems.

Literature

Landau, R.: "The concept of the male climatic". Med. Clin. N. Amer., 35:279-288, 1951.

Ryan, R.J.: "Uncertain and hypothetic disorders; unclassifiable syndromes." D.M., 27-36 (March), 1961.

COCHRANE SYNDROME

Synonymy

Familial leucine-sensitive hypoglycemia. S. of leucinosensive hypoglycemia.

Syndromography

Clinic

Hypoglycemic crises appear in the child (see hypoglycemic syndromes in the newborn) after ingestion of proteins. In adults, it is generally asymptomatic; Ingestion of a test dose of leucine or prolonged fasting induces hypoglycemic crisis.

Paraclinical examinations

Blood. Fasting hypoglycemia (very common), postprandial hypoglycemia. Leucine tolerance test.

Syndromogenesis and etiology

In adults: idiopathic, familial, associated with adenoma of the islet cells of the pancreas. It can also occur after treatment with chlorpropamide.

The etiology in children is unknown.

Literature

Cochrane, W.A.; W.W. Payne: M.J. Simpkiss, et al.: "Familial hypoglycemia precipitated by amino acids." J. Clin. Invest., 35:411-422, 1956.

MYXEDEMATOUS COMA SYNDROME

Syndromography

Clinic

Predominates in females. In hypothyroid patients, its precipitating cause is generally an infection, other times anesthesia or drugs.

Physical examination. Hypothermia, hypotension, heart failure with progression to shock and coma.

Paraclinical examinations

Blood. Low sodium and chlorine.

Urine. Low 17-ketosteroids.

Generally there is no time for investigations due to the emergency of giving treatment.

Syndromogenesis and etiology

Inability to cope with stress in patients with a significant thyroid hormone deficiency. Adrenal insufficiency.

Literature

Catz, B. and S. Russell: Myxoedema shock and coma". Arch. Int. Med., 108:407-4 17, 1961.

LeMarquand, H.S.; W. Hausmann and E.H. Hemsted: Myxoedema as a and cause of death: report of two cases." Br. Med. J., 1:704-706, 1953.

CONDORELLI SYNDROME

Synonymy

S. venous obesity.

Syndromography

Clinic

Obese patients, slight facial cyanosis that increases when leaning, episodes of tonic-clonic muscle contractions, altered sleep rhythm, altered respiratory rhythm and frequency in the recumbent position.

Physical examination. Diffuse obesity, no edema, facial cyanosis, turgor without pulsation in external jugular veins, spasm of retinal vessels.

Paraclinical examinations

venous pressure. Augmented (diagnosis).

Chest radiology.

Electrocardiogram and electroencephalogram.

Syndromogenesis and etiology

Fatty infiltration of the mediastinum and salt and water retention constitute the mechanisms that explain the symptoms.

Literature

Condorelli, L.: "Clinical pathophysiology of the mediastinum (sistezione su base anaatomo-fisiologica delle sindromi mediastiniche sense strictiori)". Cong. Soc. Ital. Med. Int., 48:1-163, 1947.

Marrazza, P. and E. Zilli: *La syndrome venosa degli adiposi di Condorelli, contributo casistico". Policlinico (Sez. Part.), 61:12-22, 1954.

PERSISTENT MÜLLER DUCT SYNDROME

Syndromography

Clínic

Rudimentary uterus and fallopian tubes, male Wolffian ducts, male spermatogenesis, normal serogenital sinus, normal hands, normal external genitalia. Male production of testosterone and estrogen.

Paraclinical examinations

Karyotype. 46,XY.

Syndromogenesis and etiology

Hereditary male pseudohermaphroditism with defects in Müllerian regression. It is transmitted by autosomal recessive inheritance.

It is due to a defect in anti-Müllerian hormone (AMH), produced by Sertoli cells. The production of this hormone requires the presence of the testicular determining factor gene found on the Yp chromosome.

CONN SYNDROME

Synonymy

S. of primary aldosteronism. S. of the electrocurtain.

Syndromography

Clinic

Frontal or central headache, polyuria greater than 2.5 L in 50% of cases. Polydipsia, nocturia or diurnal diuresis during daytime bed rest, initial muscle weakness, with long-lasting transient paralysis and tetanic crises.

Physical examination. Moderate or intense non-malignant arterial hypertension. Fundus: tortuous arteries. Osteotendinous hyporeflexia.

Hyperventilation and the Trousseau maneuver produce tetanic crises.

Paraclinical examinations

Blood. Alteration of electrolyte balance:

hypokalemia, the reduction of sodium in the diet causes a increased serum potassium; In advanced cases there is hypernatremia and increased CO2.

Urine. Increased excretion of potassium and aldosterone.

Decreased plasma renin. With the administration of spironolactone, sodium excretion increases and potassium excretion decreases. The administration of thiazides or another diuretic early reveals the tendency to potassium loss.

Hypocalcemia.

Electrocardiogram. Inverted T waves, prolonged ST interval, improve with spironolactone or potassium.

Radiography. Retrograde phlebogram of the peripheral adrenal veins. Arteriography shows an avascular area with an opacity corresponding to the tumor.

Laboratory exams. The test with administration of a thiazide diuretic induces muscle cramps and weakness.

Potassium dosage. Altered glucose tolerance test, corrected by the administration of potassium.

Slight increase in serum and/or urinary aldosterone with high salt intake, normal excretion of 17-hydroxycorticoids.

Cortisone administration test. If there is a tumor, adrenal secretion of aldosterone continues. If it is due to hyperplasia, hypersecretion is suppressed.

Syndromogenesis and etiology

Hyperplasia or tumor of the adrenal cortex causes an overproduction of aldosterone, a steroid hormone that acts on electrolytes. The same symptoms can be produced by other causes that increase the production of aldosterone, as observed in the so-called secondary aldosteronisms.

Literature

Conn, J.W.: "Primary aldosteronism: a new clinical syndrome." J. Lab. Clin. Med., 45:3-17, 1955.

Conn, J.W.: "Aldosteronism in men." J.A.M.A., 183:871-878, 1963.

Conn, J.W.; HE. Cohen and D.R. Rovner: *Normokalemic primary aldosteronism, a detectable cause of essential hypertension":. J.A.M.A., 193:200-206, 1965.

CUSHING SYNDROME

Synonymy

S. of hypercorticism.

Syndromography

Clinic

Three clinical forms are recognized:

1. Cushing syndrome due to adrenal neoplasms.
2. Cushing's disease.
3. Ectopic ACTH syndrome.

CUSHING SYNDROME DUE TO ADRENAL NEOPLASMS

Syndromography

Clinic

It is characterized by: weakness, fatigue, oligomenorrhea, back pain, loss of libido, impotence in men, mental disorders (from simple irritability to schizophrenia).

Physical examination. Obesity of the face, neck ("buffalo nape") and trunk, plethoric facies ("moon face") with pink-carmine cheeks, purpuric striae on the abdomen and lower limbs, ecchymosis, peripheral edema, hirsutism, hypertension, cardiomegaly.

Paraclinical examinations

Urine. Glucosuria, increased 17-ketosteroids and 17-hydroxycorticoids.

Blood. Decreased ACTH, increased 17-hydroxy-corticosteroids, decreased eosinophils and lymphocytes, increased neutrophils and red blood cells. Insulin-resistant hyperglycemia, hypercholesterolemia.

ACTH stimulation. Lack of response if the tumor is malignant, normal if it is a benign adenoma.

Response to methopyrone. Insufficient, normal in benign adenoma.

Suppression with dexamethasone. Lack of response, normal in benign adenoma.

Radiology. Osteoporosis, nephrocalcinosis. Pathological fractures with the formation of large bone calluses. Aseptic necrosis of the epiphyses. Vertebral crushing.

Urogram. An adrenal mass may be seen and is best defined with computed tomography.

Arteriography. Shows the tumor.

Computerized axial tomography. Show the turn.

Syndromogenesis and etiology

This syndrome is caused by adrenal neoplasia with autonomous secretion of cortisol (not dependent on ACTH). This results in suppression of ACTH production and atrophy of normal adrenal tissue. Neoplasms can be benign or malignant and produce the syndrome whenever they are hypersecretory.

It can also be caused by disorders of hypothalamic function, adrenocortical hyperplasia, non-adrenal or pituitary tumors with ectopic production of ACTH, or ACTH-like (pulmonary and prostatic neoplasms).

In the literature, the name Cushing's disease is reserved for basophilic adenomas of the pituitary gland.

The most common cause of Cushing's syndrome is iatrogenesis caused by prolonged and inappropriate administration of adrenal steroids.

CUSHING'S DISEASE

Syndromography

The clinical picture is the same as that described in Cushing's syndrome due to adrenal neoplasms.

Paraclinical examinations

Blood. Increased ACTH or prolonged secretion during the day; increased 17-hydroxycorticoids. Hematological and biochemical alterations (see Cushing syndrome due to adrenal neoplasms).

Urine. See Cushing's syndrome.

ACTH stimulation test. Three- to five-fold increase in urinary 17-hydrocorticoids.

Response to methopyrone. Qualitatively normal.

Suppression with dexamethasone. There is a response only with high doses.

Computerized axial tomography. Turkish chair enlarged in size.

Syndromogenesis and etiology

Inappropriate levels of ACTH produced by basophilic tumors of the pituitary gland resulting in adrenal hyperplasia with increased production of corticosteroids.

Literature

Cushing. H.: "The basophil adenomas of the pituitary body and their clinical manifestations (pituitary basophilism)." Bull. Johns Hopkins Hosp., 50:137-195, 1932.

ECTOPIC ACTH SYNDROME

See Cushing's syndrome.

Syndromography

Clinic

Weakness, fatigue, oligomenorrhea in women. Symptoms due to ACTH produced by the tumor (lung, pancreas).

Physical examination. Ecchymosis, edema. Many of the signs of Cushing's syndrome do not occur due to the rapid course of the disease; typical obesity usually does not develop, hypertension is observed only in some cases.

Paraclinical examinations

Blood. High ACTH, high cortisol.

Urine. increased 17-ketosteroids and 17-hydroxycorticosteroids; Other typical changes are due to excessive cortisol production and changes due to the primary tumor.

ACTH stimulation. Variable response according to circulating ACTH levels.

Response to mepirapone. Variable according to circulating cortisol levels.

Suppression with dexamethasone. Negative response even with high doses.

Syndromogenesis and etiology

It is due to the production of a protein substance similar to ACTH by tumors: of the lung, most frequently, almost always oat cell carcinoma, pancreas, breast, prostate, parathyroid, thyroid, ovaries, sympathoblastoma and thymus.

ACTH-like substance induces adrenal hyperplasia.

It is considered among the paraneoplastic syndromes.

Literature

Brown, W.H.: “A case of pluriglandular syndrome.” Lancet, 2:1022-1023. 1928.

Liddle, G.W.; J.R. Givens; W.E. Nicholson, et al.: "The ectopic ACTH Syndrome." Cancer Res. 25:1057-1061, 1965.

Strott, C.A.; AC Nugent and F.H. Tyler: "Cushing's syndrome caused by bronchial adenomas." Amer. J. Med., 44:97-104, 1968.

TONI-DEBRÉ-FANCONI SYNDROME

Synonymy

S. by De Toni-Fanconi. S. de Fanconi-De Toni-Debre. S. of hypochloremic osteonephropathy. S. of glucosuric-hypochloremic osteonephropathy. S. of aminodiabetes. S. of kidney rickets-

phosphoglyco-aminoaciduria. Fanconi II disease. S. of nephrotic-glucosuric dwarfism with hypophosphatemic rickets. S. of osteomolacia-renal glucosuria-aminoaciduria--hyperphosphaturia. S. of cystinuria I.

Syndromography

Clinic

This syndrome corresponds to different etiologies and, according to them, presents different characters depending on its cause.

There are two ways.

Child form:

It affects both sexes. There is normality at birth and in the postnatal period. It generally begins at 4 or 6 months of age. Patients present insufficient development, vomiting, unexplained fever, polyuria, dehydration, polydipsia, anorexia, photophobia. Dwarfism and hypophosphatemic rickets are usually present, and congenital defects may exist.

Adult form:

Osteomalacia and "pseudofractures", ecchymoses and hemorrhages, as well as brownish pigmentation of the skin due to hemosiderosis, sometimes

There may be weakness (due to loss of potassium), and dehydration (due to loss of water).

Sometimes diseases are associated with the clinical picture.

malignant, hemolymphopoietic diseases such as leukemia and multiple myeloma.

These and other clinical manifestations are related to the biochemical alterations and the various etiologies that can produce them. Renal failure is rare and the prognosis is good when systemic manifestations are treated.

Paraclinical examinations

Blood. Anemia, thrombocytopenia and leukopenia. Hypochloremic acidosis, hyperaminoacidemia. Hypercalcemia, normal glucose. Thrombopenia, due to bone marrow hypoplasia. Sometimes leukemia or multiple myeloma. Normal glucose. At the beginning, low levels of phosphates that gradually become normal; normal calcium, elevated alkaline phosphatase, hypokalemia.

Urine. Hyperphosphaturia, hyperaminoaciduria, hypercalciuria, glycosuria (without hyperglycemia), bicarbonate loss (causing acidosis).

Radiology. Osteomalacia and sometimes pseudofractures can be observed.

Kidney biopsy. There is usually a "swan-neck" deformity in the initial portion of the proximal tubule, which is possibly the anatomical basis of the tubular alteration. It is debated whether it is secondary to the functional deficit.

Cystine dosage. It can be performed on bone marrow, kidney, leukocytes and conea.

Culture of fibroblasts in amniotic fluid.

Syndromogenesis and etiology

It is a congenital defect that is characterized by an alteration of protein metabolism. There is a functional abnormality of the proximal convoluted renal tubules.

The syndrome can be congenital, due to autosomal recessive inheritance with multiple allelic mutations and alterations in the locci, which determines the various forms of the syndrome.

It can also be acquired after conditions that have a genetic basis: cystinosis, tyrosinosis, galactosemia, fructose intolerance, glycogen storage disease (type I), Wilson's disease, familial nephrosis, etc. In Occasionally it is triggered by poisoning by: cadmium, lead, uranium, tetracyclines.

Lowe syndrome (oculo-cerebro-renal) is linked to a recessive character related to the X chromosome typical of Fanconi syndrome and is accompanied by ocular and brain abnormalities.

There are idiopathic forms.

The common denominator is a renal tubular injury with an anatomical and functional disorder of the excretion of amino acids, glucose and phosphate.

The decreased capacity to transfer water and solutes across the tubular cell membrane appears to be the main mechanism of the alterations of this syndrome.

Literature

Fanconi, G.: "Die nicht diabeticischen Glykosurien und Hyperglykamien des alteren Klndes." Jahrb. Kinderh., 133:257- -300. 1931

De Toni, G: “Remarks on the relations between renal rickets (renal dwarfism) and renal diabetes.” Acta Paediatr., 16:479-484,1933.

Salassa, R.M. et al.: "Observations on metabolic effects of vitamin D in Fanconi' s syndrome." Proc. Staff Meet. Mayo Clin., 29:214, 1954.

DE MORSIERI SYNDROME

Synonymy

S. of olfactogenital dysplasia in the female.

Syndromography

Clinic

It only affects females and is rare.

Physical examination. Anosmia, ovarian hypoplasia, and therefore underdevelopment of secondary sexual characteristics.

Syndromogenesis and etiology

Hypoplasia is most likely related to the receptivity of the ovaries that do not respond to stimulation of the anterohypophysis (genetic defect?) although it continues its normal secretion. Under certain circumstances the ovaries are capable of reacting to exogenous stimulation in the form of human gonadotropin extracted from the urine of menopausal women. From a practical point of view, emphasis should be placed on the importance of a rhinological examination in case of severe ovarian hypoplasia and also a gynecological examination in women with anosmia in whom this disorder may be of central origin.

Literature

De Morsier, G.: *Median cranioencephalic dysraphias and olfacto-genital dysplasia". World Neurol., 3:485-506, 1962.

Muller, P.; G Geiner and A. Wackenheim: "La dysplasie olfacto- genitale ou syndrome de De Morsier, Etude Clinique, discussion, pathogenique, problems etiologiques et therapeutiques." Strasbourg. Rev. Oto-Neuro-Ophtal, 38/6, 273-286, 1966.

ISOLATED GROWTH HORMONE DEFICIENCY SYNDROME

Synonymy

S. of idiopathic pituitary dwarfism. S. of atellosis. S. of pituitary dwarfism.

Syndromography

Clinic

It affects both sexes, gestational age and birth weight are generally normal. Growth during the first year is usually normal and then severely retarded.
Sometimes there is a late onset around age 10 with slow growth. Normal or delayed pubertal development, height more delayed than weight.

Paraclinical examinations

Plasma. Absent or decreased growth hormone.
no increase after insulin stimulation. Normal levels of TSH, ACTH and gonadotropins, hypoglycemia.
Radiology. Marked delay in bone age. No injuries sellars.

Syndromogenesis and etiology

Isolated deficiency of growth hormone secretion. Familial cases that are transmitted by autosomal recessive inheritance have been reported but are rare; They are often due to craniopharyngiomas and other non-genetic causes.
Patients show sequential loss of trophic hormones from the anterior pituitary.

Literature

Antonin, J.M.E.: "Hypothalamo-hypophysarer Zwergwuhs mit spontaner." Puberty. Helvet. Paediat. Acta., 16:267-276, 1961.
Gilford, H.: “Ateleiosis: form of dwarfism.” Practitioner, 70:797-819, 1903.

McArthur, R.G. et al.: "The natural history of familial hypopituitarism." Am. J. Med. Genet., 22:553-566, 1985.

5-ALPHA-DEFICIENCY SYNDROME

-REDUCTASE

Syndromography

Clinic

1. Severe perineoscrotal hypopospadias, hooded foreskin, ventral urethral cleft, and urethral opening at the base of the penis.
2. Blind vaginal pouch of variable size that opens into the urogenital sinus or urethra, immediately behind the orifice.
urethral cyst.
3. Well-developed testicles, with normal epididymides, vas deferens and seminal vesicles, opening of the ejaculatory ducts into the blind-bottomed vagina.
4. Female external habit without breast development but with normal axillary and pubic hairs.
5. Absence of internal female genitalia.
6. Masculinization to a variable degree upon reaching puberty.

Paraclinical examinations

Plasma testosterone. Normal.
plasma LH. Little elevated; deficiency of the enzyme 5- -alpha-reductase measured in tissue biopsy and cultured fibroblasts; Other times this enzyme is functionally absent or its structure is abnormal although it is synthesized normally.

Syndromogenesis and etiology

Autosomal recessive form of male pseudohermaphroditism. In a male embryo with normal synthesis of Testosterone and normal androgen receptors, a failure in the production of dihydrotestosterone would be expected to result in this phenotype, due to deficiency of the enzyme 5-reductase, which converts testosterone to dihydrotestosterone.

CYCLOHYDROLASE DEFICIENCY SYNDROME

Syndromography

Clínic

It affects both sexes, and manifests itself from birth.
Physical examination. Mental retardation, microcephaly, erratic eye movement, bilateral optic atrophy, normal reflexes.

Paraclinical examinations

Electroencephalogram. Hypsoarrhythmia.
Computerized axial tomography. It shows dilation of the cerebral ventricles, cortical atrophy.
Blood. Decreased cyclohydrolase activity in erythrocytes and liver tissue. No decrease in forminotransferase activity in erythrocytes and liver tissue.

Syndromogenesis and etiology

Cyclohydrolase deficiency.

Literature

Arakawa, T.S.: M. Fujii: K. Ohara; S. Watanabe, et al.: "Mental retardation with hyperfolic-acidemia not associated with formiminoglutamic aciduria; cyclohydrolase deficiency syndrome." Tohoku J. Exp. Med., 88:341-352, 1966.

TRIOSPHOSPHATE ISOMERASE DEFICIENCY SYNDROME

Synonymy

S. of TPI (triosephosphate isomerase) deficiency.

Syndromography

Clinic

It affects both sexes, begins in childhood. Chronic hemolysis or hemolytic crisis. In homozygotes, after the first year of life, a progressive neurological atypical syndrome appears, splenomegaly, recurrent infections, and sudden death.

Paraclinical examinations

Blood. TPI deficiency in red blood cells and leukocytes, slight macrocytosis, some red blood cells with digital extensions. Autohemolysis is almost always completely corrected with glucose or adenosine triphosphate.

Syndromogenesis and etiology

Triosephosphate isomerase deficiency in red blood cells and leukocytes.

Literature

Schneider, A.S.; W.N. Valentine; M. Hattori, et al.: "Herediatry hemolytic anemia with triosephosphate isomerase deficiency." N. Engl. J. Med., 272:229-235, 1965.

MULTIPLE PITUITARY DEFICIENCY SYNDROME

Synonymy

S. de Burnier. (See pituitary dwarfism.)

Syndromography

Clinic

It affects both sexes, age of onset varies, the characteristics are diverse according to the number of hormones in deficit. All cases present growth retardation; symptomatic hypoglycemia is another ever-present fact;

Hypothyroidism, hypogonadism and hypoadrenalism appear variably.

Paraclinical examinations

Blood. Determination of growth hormone, ACTH, TSH, gonadotropins; hypoglycemia. another disorder metabolic according to the hormone involved.

Syndromogenesis and etiology

The syndrome may be of idiopathic cause or secondary to an adenoma. It can also be caused by a destructive lesion of the pituitary gland or a diencephalic pathology.

Literature

Burnier, R.: “A new hypophyseal syndrome; hypophyseal nanism." Ann. Ophthal., 21:263-273, 1912.

Gilford, H.: "Ateleiosis: from of dwarfism." Practitioner, 70:797-819, 1903.

Rabkin, M.T. and A.G. Frantz: “Hypopituitarism: A study of growth hormone and other endocrine functions.” Ann. Int. Med., 64:1197-1207, 1966.

POLYGLANDULAR DEFICIENCY SYNDROME

Synonymy

Schmidt's S.

Syndromography

Clinic

The first sign appears in adult life. It encompasses any combination of adrenal insufficiency, lymphocytic thyroiditis, hypoparathyroidism, and gonadal insufficiency, often also saccharine

Physical examination. The manifestations can be so extensive that they simulate panhypopituitarism.

Paraclinical examinations

Blood. Low concentrations of circulating hormones; antibodies against gastric parietal cells and eventually achlorhydria; Antibody-mediated IgA deficiency may be found.

Syndromogenesis and etiology

Most cases are sporadic and female; Several members of a family have been affected and those without overt disorder show serologic abnormalities indicative of immune disorder, suggesting a genetic basis. A relationship has been found with HLA-B8 and DW3 antigens in whites and BW5 in Japanese with associated hyperthyroidism. An association has also been found with other immune disorders such as sprue, vitiligo, myasthenia gravis, pernicious anemia, and aplastic anemia. The basic lesion may lie in a mutation of some inherited immune mechanism, perhaps it is the heterogeneous cause.

BICARBONATE DEFICIT SYNDROME

Synonymy

S. of metabolic alkalosis.

Syndromography

Clínica

Weakness, malaise, headache, nausea, vomiting, abdominal pain, Kussmaul's breathing. If the condition persists, respiratory depression occurs. There are associated manifestations of the underlying disease: diabetes, uremia, congestive heart failure.

Paraclinical examinations

Blood. Leukocytosis.

1. Decompensated metabolic acidosis: low pH, pCO, normal, low bicarbonate, high chlorine, low sodium, normal or high depending on the cause.
2. Compensated metabolic acidosis: low or normal pH, low pCO, low bicarbonate, low normal or high chlorine, low or normal sodium, high potassium.

Urine. Acidic with increased ammonia.

Syndromogenesis and etiology

It is due to an excess of organic or inorganic acids due to:

1. Excessive production (diabetes, wasting, high fever, hyperthyroidism, violent exercise, shock).
2. Ingestion (sodium, calcium or potassium chloride, carbonic anhydrase inhibitors, cation exchange resins, salicylate poisoning, methyl alcohol poisoning).
3. Retention (kidney diseases).
4. Due to loss of bases (diarrhea, fistulas, ureteroenterostomies, Light Wood syndrome (see this one).

Literature

Goldberger, E.: A Primer of Water, Electrolyte, and Acid-base Syndromes. ed. 3, Philadelphia, Lea & Febiger, 1965.

DEL CASTILLO SYNDROME

Synonymy

S. of testicular dysgenesis. S. of germ cell aplasia.

Syndromography

Clínic

Sterility with normal libido and erection.

Physical examination. Normally developed men with all secondary sexual characteristics, small or normal testicles.

Paraclinical examinations

Spermogram. Decreased 17-ketosteroids, FSH normal.

Syndromogenesis and etiology

The syndrome is caused by a developmental defect (similar to that seen after radiation exposure).

Literature

Del Castillo, E.: A. Trabuco and F.A. de la Balze: Syndrome produced by absence of germinal epithelium without impairment of Sertoli or Leydig cells". J. Clin. Endocrinol., 7:493-502, 1947.

PHOSPHORUS DEPLETION SYNDROME

Syndromography

Clínic

Patients receiving prolonged treatment with non-absorbable antacids, such as aluminum and magnesium hydroxide, in antiulcer therapy, in the prevention of kidney stones, etc. Weakness, anorexia, malaise. In the severe form, bone pain and joint stiffness appear, occasionally intentional tremor.

Paraclinical examinations

Urine. Hypophosphaturia, hypercalciuria.

Blood. Hypophosphatemia. Increased gastrointestinal calcium absorption.

Radiology. Osteomalacia.

Syndromogenesis and etiology

Phosphorus depletion as a consequence of prolonged administration of antacids.

Literature

Bloom, W.L. and D. Finchum: "Osteomalacia with pseudofractures caused by ingestion of aluminum hydroxide." J.A.M.A., 174:1327-1330, 1960.

Lotz, M.E. Zisman and F.C. Bartter: *Evidence for a phosphorus-depletion syndrome in man". N. Engl. J. Med., 278:409-415, 1968.

PURE WATER DEPLETION SYNDROME

Synonymy

S. of water loss. S. of pure dehydration. S. of hypertonic dehydration. S. of water deficit. S. de Leutscher.

Syndromography

Clinic

Thirst (may be absent in patients with brain injuries); inability to swallow dry food, weakness, weight loss, fever, personality changes, hallucinations, delirium.

Physical examination. Dry and hot skin, xerostomia, absence of lacrimation, dry and cracked tongue, skin fold, tachycardia, hyperpnea and coma (terminal signs).

Paraclinical examinations

Blood. At the beginning there are no alterations, when hemoconcentration is established, the hematocrit, electrolytes and proteins rise, hyperazotemia.

Urine. Scant, high density unless there is prior difficulty concentrating. Hematuria, proteinuria, cylindruria.

Syndromogenesis and etiology

This syndrome can be present in different situations: loss of water, non-ingestion (shipwrecked, patients who do not swallow, etc.), brain lesions that make it impossible to ingest water, diabetes insipidus, water-losing nephritis, diarrhea, hyperventilation, burns, polyuric syndromes.

Literature

Christie, S.B.M. and E.J. Ross: "Ectopic pinealoma with adipsia and hypernatremia." Br. Med. J., 2:669-670, 1968.

Schoolman, H.M.; A. Dubin and W.S. Hoffman: "Clinical syndromes associated with hypernatremia." Arch. Int. Med., 95:15-23, 1955.

DEHYDRATION SYNDROME

Syndromography

Clinic

Dry skin and mucous membranes, oliguria, asthenia, muscle cramps with hypotonia, ocular hypotonia and enophthalmia, fever in children.

Paraclinical examinations

Blood. Hemoconcentration, hyperazotemia (extrarenal).

Syndromogenesis and etiology

Loss of water and electrolytes due to profuse sweating, abundant diarrhea, involuntary vomiting, poor fluid intake, etc.

DIABETES INSIPIDAL SYNDROME

Syndromography

Clínica

It predominates in men; It generally begins at an early age. Polydipsia with insatiable thirst. Polyuria in the range of 15-30 L daily. Nocturia, enuresis. Dehydration, constipation.

Physical examination. Dry skin and mucous membranes.

Paraclinical examinations

Urine. Low density.

Radiology of the sella turcica.

Computerized axial tomography.

Brain scan.

Lumbar puncture.

Secretion tests. Nicotine, hypertonic saline, vasopressin.

Syndromogenesis and etiology

It is due to structural damage to the hypothalamus-neurohypophyseal axis caused by: trauma, tumor surgery,

granulomas. There are two known forms of inheritance: sex-linked recessive and autosomal dominant.

Literature

Forssman, H.: "On hereditary diabetes insipidus with special reference to a sex-linked form." Acta Med. Scand., 121, Suppl. 159, 1956.
Grinker, RR. and A.L. Sahs: Neurology. ed. 6, Springfield, Thomas, 1966.

ADH-RESISTANT DIABETES INSIPIDAL SYNDROME

Synonymy

S. diabetes insipidus resistant to pitresin. S. of nephrogenic diabetes insipidus.

Syndromography

It includes all conditions where renal tubular inability to respond to antidiuretic hormone (ADH) predominates over any other renal manifestation present.

Clínica

See diabetes insipidus syndrome.

Mental and physical retardation are observed in some patients with congenital forms. Occasionally there are symptoms and signs of dehydration. Congenital forms affect only the male (absolute absence of response to ADH). Limited response has been observed in some women from affected families.

Paraclinical examinations

See diabetes insipidus.

The administration of ADH does not modify the symptoms.

Syndromogenesis and etiology

Congenital:

1. X-linked hereditary nephrogenic diabetes insipidus syndrome with variable degree of manifestations in females.

2. Associated with Fanconi or Lightwood syndrome.

Acquired:

1. Hypokalemic syndrome.
2. Hypercalcemic syndrome.
3. Conn syndrome.
4. Chronic pyelonephritis.
5. Obstructive uropathies.
6. Hereditary syndrome linked to the X chromosome that causes serious disorders in males.

Literature

Vorherr, H.; S.G Massry; R.D. Utiger, et al.: "Antidiuretic principle in malignant tumor extracts from patients with inappropriate ADH syndrome." J. Clin. Endocr., 28:162-168, 1968.
Williams, R.H. and C. Henry: "Nephrogenic diabetes insipidus: transmitted by females and appearing during infancy in males". Ann. Int Med. 27:84-95. 1947.

DIABETES INSIPIDAL SYNDROME LIKE"

Syndromography

Clínica

1. Glucosuria.
2. Accentuation of diabetes mellitus.
3. Syndrome mimicking diabetes insipidus; In addition, polyuria, polydipsia, muscle weakness, incoordination.
4. Hyperglycemia.

Syndromogenesis and etiology

It occurs especially in neoplasms, preferably of the lung, which are capable of secreting a substance whose action is almost identical to the hormone ACTH of the pituitary gland. It is included in paraneoplastic syndromes.

DIABETES MELLITUS SYNDROME

Syndromography

Clínica

Classic triad of polyuria, polydipsia, polyphagia.

In the course of its evolution: vascular alterations (retinal such as retinopathies of different intensity), arterial (ulcers, gangrene, etc.) If the patient does not follow the treatment properly, complications are early and serious. Diabetic acidosis is a serious complication that can lead to coma and death.

Paraclinical examinations

Blood. Fasting or provoked hyperglycemia. Electrolyte alterations when decompensated.

Urine. Glucosuria. Ketonuria in diabetic acidosis crises.

Syndromogenesis and etiology

Often hereditary, possibly polygenic or multifactorial.

1. Dysfunction of the pancreas with deficient insulin secretion.
2. Inhibition of the effect of insulin.
3. Inability to store ingested carbohydrates.
4. Hypofunction or hyperfunction of certain endocrine glands (hyperfunctioning tumor of the pituitary gland, adrenal medulla or cortex, thyroid, etc.).
5. Other causes such as: therapeutic administration of cortisone, destructive liver injury, hemochromatosis, etc.
6. Genetic, autoimmune, or insulin receptor alterations.

At the moment it is classified into:

Primary:

- Insulin-dependent saccharine diabetes, type I.
- Non-insulin-dependent saccharine diabetes, type II.
- Adult non-insulin-dependent saccharine diabetes.

Secondary:

- Saccharine diabetes associated with other conditions.

Literature

Aretaeus of Cappadocia, A.D. 81-131. Diabetes Mellitus. ed. 7, Indianapolis, Lilly, 1967.
Gondos, B.: "Roentgen observation in diabetes osteopathy." Radiology, 91:6-13, 1968.
Zitomer, B.R.; H.EF. Gramm and GP. Zozak: "Gastric neuropathy in diabetes mellitus, Clinical and radiological observations." Metabolism, 17:199-211, 1968.

DIABETES SACCHARIN SYNDROME - DIABETES INSIPIDIA - OPTICAL ATROPHY

Syndromography

Clínica

It has been observed in brothers. There may also be mild neurological hearing loss. Saccharine diabetes is insulin-dependent early on and diabetes insipidus usually appears before the age of 20.

Syndromogenesis and etiology

It is probably a rare autosomal recessive defect.

DIABETIC SYNDROME

From a clinical point of view, two syndromes have been defined: acute and chronic (see both). Complications of the disease and some effects of prolonged treatment are also manifested in a number of complex symptoms according to the organ or system affected; Sometimes these manifestations appear before the disease has been diagnosed, others become clinically evident during the course of the condition.

Vascular insufficiency:

1. Large vessels affecting the brain, kidneys, heart, lower limbs, etc.

2. Small vessels, microangiopathy, retinopathy.

Neurological disorders:

1. Neuritic syndrome, diabetic amyotrophic syndrome.
2. Visceral neuropathy that affects:

- Eyes, external ophthalmoplegia.
- Gastrointestinal tract, gastric neuropathy, diarrhea.

Genitourinary tract, kidney (Kimmelstiel-Wilson syndrome), neurogenic bladder, impotence.

Neuropathic arthropathy (most common in tarsal and ankle joints).

Neurotrophic ulcers.

Autonomic nervous system, orthostatic hypotension, anhidrosis, tachycardia.

Literature

Aretheus the Cappadocia, A.D. 81-131. Diabetes Mellitus. ed, 7, Indianapolis Lilly, 1967.
Behrendt, T.; R.D. Raymond and T.D. Duane: "Evaluation of microlesions in diabetic retinopathy." Amer. J. Opthal., 64:721-725, 1967.

ACUTE DIABETIC SYNDROME

Syndromography

Clinic

It occurs more frequently in juvenile diabetes mellitus.

It presents a picture of ketosis: light nausea, thirst, general malaise, and acidosis: vomiting, dizziness, disorientation, hypernea.

Physical examination. Dryness of the skin and mucous membranes, hypotonia of the eyeballs, apple odor, Kussmaul's respiration, fever and coma.

Paraclinical examinations

Urine. Glucosuria, ketonuria.

Blood. Increased acetone, intense hyperglycemia (important to rule out hypoglycemic coma). Increased potassium and decreased sodium.

Blood culture. In case of infections.

Syndromogenesis and etiology

They are patients with known or unknown diabetes mellitus, in whom this condition is precipitated by different factors: infections, vomiting, diarrhea, circulatory insufficiency, violations of the diet or in the administration of insulin or oral hypoglycemic agents.

CHRONIC DIABETIC SYNDROME

Syndromography

Clinic

It affects both sexes and begins at any age. There is polyuria, polydipsia, polyphagia and weight loss, greater in children. Vulvar and generalized itching, premature tooth loss, asthenia, drowsiness, impotence, history of macrofetus and hydramnios.

Physical examination. Depending on the stage of the condition and the severity of the diabetic condition, the following findings may be found. Eyes: premature cataract, microaneurysms, vitreous and retinal hemorrhage. Skin: fungal infections (Candida), xanthochromia, xanthomas. Cardiovascular: atherosclerotic vascular occlusion, incurable leg ulcers with gangrene, edema and heart failure. Renal: Kimmelstiel-Wilson syndrome, nephrosclerosis, chronic pyelonephritis, necrotizing papillitis. Neurological: peripheral neuritis, areflexia, apalesthesia, neurogenic bladder, nocturnal diarrhea, coma.

Paraclinical examinations

Urine. Glucosuria, ketonuria.

Blood. Hyperglycemia, ketosis, hypercholesterolemia; hypertriglyceridemia,

Glucose tolerance test. Decreased after administering cortisone.

Antibodies to insulin. In blood and tissues.

Insulin dosage.

Cortisone test.

Retinogram.

Skin and muscle biopsy.

Radiology. Calcification of the aorta.

Syndromogenesis and etiology

It is caused by absolute or relative insulin deficiency due to: congenital or acquired defect in its production,

dysfunction of the beta cells of the islets of Langerhans, hormonal imbalance or neutralization of insulin (due to autoantibodies or acquired). Microangiopathy may be present from birth as a localized disorder or adopt a diffuse distribution.

CONGENITAL DIARRHEA WITH HYPOCHLOREMIC ALKALOSIS SYNDROME

Synonymy

S. of chlorurrea.

Syndromography

Clínica

It manifests after birth with profuse watery diarrhea without vomiting, difficulty gaining weight, and good appetite except in periods of dehydration.

Physical examination. Negative.

Paraclinical examinations

Blood. Low chlorine, high bicarbonate, low sodium, normal potassium except when diarrhea is severe; High pH (7.64). Normal creatinine, blood glucose, protein and calcium.

Stool. High chlorine, low sodium. Acid pH, normal gastric and duodenal secretion.

Urine. pH occasionally higher than plasma, no color, low sodium, variable potassium.

Syndromogenesis and etiology

Congenital defect that affects the intestinal absorption of water, chlorine and sodium.

Literature

Gamble, J.: "Congenital alkalosis with diarrhea." J. Pediat.. 26:519-532, 1945.

Milue, M.D.: "Hereditary abnormalities of intestinal absorption." Br. Med. Bull., 23:279-284, 1968.

PURE GONADAL DYSGENESIA SYNDROME (XY)

Syndromography

Clinic

Female phenotype, lack of puberty and fertility. No stigma of gonadal dysgenesis syndrome (see this one).

Paraclinical examinations

Chromosome study. XY karyotype. At the time of the early appearance of pubertal symptoms, gonadal activity is not evident and there is an increase in gonadotropins.

Syndromogenesis and etiology

Failure in the development of testicles in the fetus and it acquires a female phenotype. There are traces of gonads in the place where one would expect to find ovaries and the presence of a uterus and small tubes. It is a different syndrome from testicular feminization, Savage and gonadal dysgenesis.

These are mutations of the SRY gene in the short arms of the Y chromosome adjacent to the pseudoautosomal region. This gene encodes a protein responsible for the differentiation of the primitive gonad, the formation of a testis and the production of local tion of a protein, gene that inhibits the development of the Müllerian ducts.

Bibliography

Frasier, S.D.; R.A. Bashore and H.D. Mosier: "Gonadoblastoma associated with pure gonadal dysgenesis in monozygous twins." J. Pediat., 64:740-745, 1964.

Sternberg, W.H. and D.L. Barclay: "Familial XY gonadal dysgenesis." Lancet, 2:946, 1967.

Wermer M.H., et al.: "Molecular determinants Tibs. mammalian sex". 21:302-307, 1996.

DYSTOCIA-DYSTROPHY SYNDROME

Synonymy

S. of dystocia-dyspituitarism.

Syndromography

Clínica

Women affected by this syndrome have a squat figure, a slightly thick neck, a wide back, short thighs, and a tendency toward obesity.

They may have male distribution of hair; hands short and thick with the three middle fingers approximately the same length. Android-type bony pelvis. Short vagina with small cervix. Subfertile. Spasmodic dysmenorrhea, tendency to abortion and high incidence of eclampsia. Labor begins with the fetus's head still high and in most cases occiput posterior. There is early rupture of membranes.

Syndromogenesis and etiology

The etiology is unknown.

Bibliography

Horner. D.A.: "Bradytocia, a study based on 500 cases in the Chicago Lying-In Hospital." Surg. Gynec. Obstet.: 44:194-201, 1927.

Williams, B.: "Dystocia dystrophia syndrome." J. Obstet. Gynec. Brit. Emp., 49:412-425, 1942.

DUCHENNE DYSTROPHY SYNDROME IN FEMALE

Syndromography

Clínica

The female is only a carrier, since it has a recessive form of transmission linked to sex. The carrier state without clinical manifestations can be identified in 80% of cases by biochemical study: increase in creatine phosphokinase. Occasionally the female may present typical clinical symptoms of Duchenne syndrome (see). The course is usually moderate and slowly evolving.

Syndromogenesis and etiology

The occurrence of forms with clinical manifestations in the female is difficult to explain by the type of hereditary pattern. The following possibilities must be considered:

1. Chromosome mosaicism.

2. Possible existence of an autosomal form of transmission.

3. "Lyon hypothesis", random inactivation of the

The basic defect corresponds to the absence of the dystrophin protein and the destruction of the cytoskeleton of the muscle cell.

Literature

Pearson, C.M.; W.M. Fowler and S.W. Wright: "X-chromosome mosaicism in females with muscular dystrophy." Proc. Nat. Acad. Sci., 50:24, 1963.

MYOTONIC DYSTROPHY SYNDROME

Syndromography

Clinic

Muscular dystrophy, premature baldness, mental retardation.

Syndromogenesis and etiology

Autosomal dominant inheritance with multiple endocrine disorders: anomalies of gonadotropins, growth hormone, hypothyroidism, saccharine diabetes, and primary gonadal insufficiency.

DORMANDY SYNDROME

Synonymy

S. of intolerance to galactose and fructose. (See hereditary fructose intolerance syndrome.)

Syndromography

Clinic

It differs from hereditary fructose intolerance because patients are fond of sweets and do not experience nausea or vomiting after ingesting fructose. Coma and seizures follow ingestion of both galactose and fructose. These symptoms disappear with the administration of intravenous glucose.

Paraclinical examinations

Blood. Following the ingestion of galactose and fructose, serum microorganic glucose and phosphorus levels decrease; normal blood glucose curve; Insulin rises after glucose ingestion.

Syndromogenesis and etiology

It has a genetic etiology with autosomal recessive inheritance.

The basic defect is unknown.

Literature

Dormandy, T.L. and R.J. Porter: "Familial fructose and galactose intolerance." Lancet, 1:1189-1194, 1961.

Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The Metabolic Basis of Inherited Diseases. ed. 2. New York. McGraw-Hill, 1967.

CYCLIC EDEMA SYNDROME

Synonymy

S. of idiopathic cyclic edema.

Syndromography

Clinic

It manifests itself in women. The edema begins in the lower limbs in the standing position; In many cases it disappears when the patient lies down, in others there is no improvement and the edema becomes generalized. In the premenstrual period it worsens.

Many patients have emotional disorders, are hysterical or psychopathic.

Physical examination. Edema that improves with bed rest.

Paraclinical examinations

Those that exclude other causes of edema, diseases of the heart, kidney, liver, thyroid, veins and simple water and sodium retention.

Syndromogenesis and etiology

Possible hypersecretion of aldosterone or increased capillary permeability of the legs.

Literature

Mach, R.S.; J. Fabre; A.F. Muller, et al.: "Idiopathique par retention sodique avec hyperaldosteronurie." Bull. Mem. Soc. Med. Hop. (Paris), 71:726-732, 1955.

CHOLESTEROL EMBOLIZATION SYNDROME

Synonymy

S. of atheromatous embolism. S. of livedo reticularis and digital infarction.

Syndromography

Clínica

The syndrome is polymorphic and highly variable. Its clinical expression is reminiscent of some collagen or vascular diseases (such as periarteritis nodosa). It mainly involves: kidney (75%) causing hypertension and kidney failure; pancreas, with symptoms and signs of acute pancreatitis; central nervous system (less common), only focal symptoms; gastrointestinal system, peptic ulcer in the elderly can sometimes have this pathogenesis; circulatory system, with obstruction of several arteries, the mesenteric with acute abdominal manifestations, the arteries of the lower limbs with livedo reticularis and occlusion.

sion of the arteries of the fingers.

Syndromogenesis and etiology

Embolization of atheromatous material in various areas of the body, spontaneously or as a result of trauma or surgery.

Literature

Flory, C.M.: "Arterial occlusions produced by emboli from eroded aortic atheromatous plaques." Amer. J. Path., 21:549-565, 1945.
Kazmier, EJ.; S.G Sheps; P.E. Bernatz, et al.: "Livedo reticularis and digital infarction: a syndrome due to cholesterol embolism arising from atheromatous abdominal aortic aneurysms." Vasc. Dis.. 3:12-24. 1966.
Panum, P.L.: "Untersuchungen uber den plotzlichen Tod durch Embolie mitteslst der durch dieselbe gesetzten Unterbrechung des Blutstromes." Arch. f. path. Anat. Berl., 25:310-338, 1862:;
Die Embolic der Arterien desgrossen Kresilaufes. Ibid, 488-530.

SCORBUTE SYNDROME

Synonymy

S. of Bachelor. S. of avitaminosis C. S. of the sailor. S. of ascorbic acid deficiency.

Syndromography

Clínica

Swelling, inflamed and "spongy" gums that bleed easily, teeth loosen and lose easily, weakness, asthenia, in terminal cases hypotension, vomiting and symptoms similar to Addisonian crisis.

Physical examination. Paleness, slight jaundice, perifollicular hemorrhages in hyperkeratotic hair follicles, hair breaks or curls under the hyperkeratotic plaques. Petechiae appear in hairy areas, ecchymosis at the slightest trauma. Tachycardia.

Paraclinical examinations

Blood. Low levels of vitamin C, normocytic or occasionally macrocytic anemia when associated with folic acid deficiency, reticulocytosis, leukopenia, low serum iron with high binding capacity, elevated indirect bilirubin.
Urine. Urobilinuria.
Stool. Positive occult blood.

Syndromogenesis and etiology

Vitamin C deficiency.

Literature

Ebers Papyrus (1500 B.C.) quoted by Majo, R.H.: Classic Descriptions of Disease with Biographical Sketches of the Authors. ed. 3, Springfield, Thomas, 1944.
Friedman, G.J. and N. Jolliffe: Vitamin C Malnutrition and Scurvy. In. Jolliffe, N. ed.: Clinical Nutrition, ed. 2, New York, Harper, 1962.

SPLENOMEGALY SYNDROME - RARE OR ABNORMAL HISTIOCYTOSIS

Syndromography

Clinic

This syndrome is asymptomatic or with a tendency to hemorrhage.
Physical examination. Great splenomegaly and hepatomegaly are detected.

Paraclinical examinations

Blood. Moderate anemia. In some cases thrombocytopenia.

Bone marrow. Hypercellular. Presence of large histiocytes, 15-20 microns in diameter, granulated (paraminosalicylic acid) positive. Morphologically the granules are different from those found in Nieman-Pick and Gaucher syndromes. It seems that the granules are made up of glycolipids and/or phospholipids.

Syndromogenesis and etiology

There is no evidence that it has a family or genetic basis.

The etiology is unknown.

Literature

Sawitsky, A.; G.A. Hyman and J.B. Hyman: "An identified reticuloendothelial cell in bone marrow and spleen: report of 2 cases with histochemical studies." Blood, 9:977-985, 1964.
Silverstein. M.N.; D.G Young: W.H. Remine, et al.: "Splenomegaly with rare morphology locally distinct histiocyte, a new syndrome." Arch. Int. Med., 114:251-257, 1964.

FAMILIAR HYPOGONADOTROPIC EUNUCODISM SYNDROME

Synonymy

S. of familial hypogonadotropic eunuchoidism. (See Launois syndrome and anosmia-eunuchoidism syndrome).

Syndromography

Clínica

It is more common in twins. When they are adolescents they have eunuchoid proportions with the absence of axillary and pubic hair, and infantile genitalia.

Paraclinical examinations

Blood. Absence of gonadotropin excretion.

Syndromogenesis and etiology

There are three reported types of inheritance of hypogonadotropic hypogonadism with anosmia, also known as Kallman.

Selective deficiency of gonadotropin secretion.

Literature

Biden, R.L. and S. Gordman: "Familial hypogonadotropic eunochoidism." J. Clin. Endocr., 15:931-942, 1955.
Huxthal, L.M.: "Sublingual use of testosterone in 7 cases of hypogonadism." J. Clin. Endocr., 3:551-556, 1943.

FAVISM SYNDROME

Syndromography

Clinic

It affects both sexes and occurs at any age. In sensitive people (in Sardinia, Sicily, Calabria and in descendants of the inhabitants of these regions). It is characterized by an acute hemolytic crisis 5-24 hours after ingesting broad beans (especially fresh). If the dust is inhaled, the reaction occurs within a few minutes and its main symptoms are:
nausea, vomiting, dizziness and fever.

Paraclinical examinations

Blood. Demonstration of sensitivity. Determination of glucose-6-phosphate dehydrogenase. Anemia, reticulocytosis and leukocytosis.

Syndromogenesis and etiology

Inherited intrinsic defect of red blood cells (related to glucose-6-phosphate dehydrogenase deficiency syndrome), plus an extrinsic toxic factor. An immune or other mechanism may be involved.

The bean contains DOPA-quinone that hemolyzes red blood cells deficient in glucose-6-phosphate dehydrogenase with the help of the genetic factor.

Inheritance is considered dominant and X-linked.

Literature

LaGrega (1856) quoted by Luisada, A.: Favism. Medicine, 20:229-250, 1941.

Wintrobe, M.M.: Clinical Hematology, ed. 6, Philadelphia, Lea & Febiger, 1967.

TESTICULAR FEMINIZATION SYNDROME

Synonymy

S. of Morris. Goldberg-Maxwell-Morris S. S. of male pseudohermaphroditism. (See Lubs syndrome.) S. of intersexuality. S. of androgynoidism.

Syndromography

Clinic

In its full clinical form, it begins in childhood and patients are raised as girls.

Physical examination. Normal external habit, they begin to develop feminine manifestations such as body silhouette, distribution of body hair, voice and breast development. They excel psychologically and socially as females.

The vagina with a blind bottom 5 cm deep can develop and the external sexual characteristics are feminine, however there may be in some cases an ambiguous growth of the penis and scarcity of pubic hair. They are attractive but sterile. Bilateral inguinal hernia, palpable testicles. Big hands and feet. Amenorrhea. Absence of uterus. Intra-abdominal gonads or along the inguinal canal.

At puberty, eunochoid growth, they reach 165 cm in height.

In the rarer incomplete form, the genitals are ambiguous, the penis is hypoplastic with hypospadias, breast development is minimal or absent at puberty, they are raised as boys and may have psychosexual confusion.

Paraclinical examinations

Blood. Normal testosterone levels. XY chromosome pattern. Absent sexual chromatin.

Syndromogenesis and etiology

Two basic defects of genetic etiology are proposed, both of which affect the androgen receptors of the cytosol: deficiency of the receptor for dihydrotestosterone (DHT) and binding defects of the receptor-DHT complex.

The gene that determines the androgen receptor is located between Xql13 and Xpl1, so an X-linked inheritance has been proposed for this defect.

Clinically, two forms of expression are proposed: the incomplete form described by Morris in 1953 and the complete form with absence or functional incapacity of the androgen receptor.
In the incomplete form there may be a certain degree of virilization of the external genitalia during puberty.

Bibliography

Goldberg, M.B. and A.FMax well: "Male pseudohermaphroditism proven by surgical exploration and microscopic examination. A case report with speculations concerning pathogenesis." J. Clin. Endocrinol., 8:367-379, 1948.

Morris, J.M.: "The syndrome of testicular feminization in male pseudohermaphrodites." Amer. J. Obstet. Gynecol., 65:1192- -1211, 1953.

Paschkis, K.E.; A.E. Rakoff; A. Cantarow, et al.: Clinical Endocrinology. ed. 3, New York, Harper & Row, 1967.

Tilley, W.D., et al.: "Characterization and expression of a cDNA encoding the human receptor." Prc. Nat. Acad. Sci., 86:327- -331, 1989.

COMPLETE TESTICULAR FEMINIZATION SYNDROME

Syndromography

Clinic

It is the most common form of male pseudohermaphroditism. Its frequency is estimated between one in 20,000 and one in 64,000 male births. It is also the third cause in order of frequency of primary amenorrhea in phenotypic women, after gonadal dysgenesis and vaginal agenesis.

Phenotypically they do not have: Müllerian and Wolffian ducts, spermatogenesis, urogenital sinus and female external genitalia. female breasts. The size is usually high; bone age and intelligence are normal. Psychosexual development is unmistakably feminine in behavior, appearance, and maternal instincts.

Paraclinical examinations

Blood. High production of testosterone, estrogen and LH.

Syndromogenesis and etiology

Recessive inheritance linked to the X chromosome. Pseudohermaphroditism linked to the X chromosome. Male pseudohermaphroditism due to a defect in the action of androgens with receptor disorders.

INCOMPLETE TESTICULAR FEMINIZATION SYNDROME

Syndromography

Clinic

This syndrome has a frequency of 10% compared to the complete form. Phenotypically, there is an absence of Müllerian ducts and spermatogenesis with male Wolffian ducts, female urogenital sinus and external genitalia with clitorimegaly and partial fusion of the labial-crotal folds.
Female breasts. The vagina is short and blind.

Paraclinical examinations

Blood. High production of testosterone, estrogen and LH.

Syndromogenesis and etiology

Recessive inheritance linked to the X chromosome. Male pseudohermaphroditism due to a defect in the action of androgens with receptor disorder.

ETHIOCHOLANOLONE FEVER SYNDROME

Syndromography

Clínic

Patients present periodic fever attacks. This syndrome can be subdivided into three groups:

1. Group with adrenogenital syndrome (see this one).
2. Group with familial Mediterranean fever-like syndrome.
3. Miscellaneous group with hypothalamic damage, impaired liver function (Hodgkin's disease, cirrhosis).

Paraclinical examinations

Plasma determination of etiocholanolone. Liver function tests and study of adrenogenital and Hodgkin syndromes.

Syndromogenesis and etiology

Tendency toward excessive production of etiocholanolone and delayed plasma clearance of unconjugated etiocholanolone.

Literature

Bondy, P.K.: G.L. Chon; C. Castiglione: "Etiocholanolone fever; a clinical study." Trans. Ass. Am. Physicians, 73:186-196, 1960.
Cohn, GL.: Periodic fever. In Beeson, P.B.. and W. McDermott. ed.: Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia. Saunders, 1967.

FORBES-ALBRIGHT SYNDROME

Syndromography

Clínic

Amenorrhea and galactorrhea associated or not with pregnancy, plus a neurological syndrome due to pituitary tumor.

Syndromogenesis and etiology

Pituitary tumor.

Bibliography

Forbes, A.P.; P.H. Heuneman; G. Griswald, et al.: “Syndrome characterized by galactorrhea, amenorrhea and low urinary FSH: comparison with acromegaly and normal lactation". J. Clin. Endocr.. 14:265-271, 1954.
Krestin, D.: "Spontaneous lactation associated with enlargement of pituitary, with report of 2 cases." Lancet, 1:928-930, 1932.
Thompson, J.P. and R.D. Kempers,: “Amenorrhea and galactorrhea". Amer. J. Obst. Gynecol., 93:65-71, 1965.

FRÖHLICH SYNDROME

Synonymy

S. of adiposogenital dystrophy. S. of sexual infantilism. S. Of hypothalamic infantilism with obesity. S. de Fröhlich- -Babinski. S. de Babinski-Fröhlich. S. de Lenois-Cléret.

Syndromography

Clínic

It is more common in men. It is manifested by delayed growth and sexual development, mental retardation, visual disorders, polyuria and polydipsia.

Physical examination. Prepubertal adiposity of the breasts, abdomen, femoral region, wide hips. Delay in the appearance of secondary sexual characteristics, pubic, axillary, facial hair. Pigmentation of the skin, it remains delicate. Dysonychia.

Paraclinical examinations

Urine. Determination of FSH, LH and 17-ketosteroids.

Vasopressin test. If diabetes insipidus coexists or is suspected.

Skull radiology. Suprasellar calcification or destructive lesion. Delay in ossification. bone growth.

In some cases there are manifestations of tumor in the hypothalamic-pituitary

Syndromogenesis and etiology

Hypopituitarism, hypothalamic dysfunction, idiopathic, tumor that compresses or envelops the anterior pituitary. Wilkins suggests reserving the diagnosis of Fröhlich syndrome only for cases where a pituitary disorder can be proven.

Literature

Bruch, H.: "The Fröhlich syndrome; report of the original case." Amer. J. Dis. Child., 58:1282-1289, 1939.
Frölich, A.: "Ein Fall von Tumor der Hypophysis Cerebri ohne Akromegalie." Vienna. Klin. Rundshan, 15:833-836, and 906-908, 1901.
Wilkins, L.: The diagnosis and treatment of endocrine disorders in childhood and adolescence. Springfield, Thomas, 1957.

ADIPOSO GYNANDRISM SYNDROME

Synonymy

See pseudo Cushing syndrome. Equivalent to adipose gynismus syndrome in females.

Syndromography

Clínica

It occurs in preadolescent males with a tendency toward obesity and delayed sexual maturation. Some morphological features are reminiscent of the female habit (due to the distribution of fat and sexual retardation). Some are above average size.

Paraclinical examinations

See pseudo Cushing syndrome.

Syndromogenesis and etiology

Genetic factor: endocrine changes possibly secondary to obesity.

The etiology is unknown.

Literature

Paschkis, K.E.; A.E. Rakoff; A. Cantarow, et al.: Clinical Endocrinology. New York, Harper Row, 1967.

Simpson, S.L.: "Clinical and pathological aspects of afrenal glands". Proc. Roy. Soc. Med., 27:383-387, 1934.

Simpson, S.L.: "Adrenal hyperfunction and function" York Acad. Bull. New Med. 27:723-742, 1951.

FAMILIAR GYNECOMASTIA SYNDROME

Synonymy

Gilbert-Dreyfus S. S. of gynecomastia-hypospadia.

Syndromography

Clinic

It affects males at pubertal age.

Physical examination. Gynecomastia, small penis, normal testicles, high-pitched voice, sparse beard, small prostate, feminoid development of the pelvis, female distribution of the fat.

Paraclinical examinations

Blood. normal 17-ketosteroids; low testosterone, high estriol, low gonadotropins. Normal karyotype.

Syndromogenesis and etiology

It is transmitted by sex-linked recessive inheritance or sex-limited autosomal dominant inheritance, suggesting a genetic etiology.

Literature

Reifenstein, E.C., Jr.: Hereditary familial hypogonadism". Rec. Prog. Horm Res., 3:224, 1948.

Rosewater, S.; G. Gwinup and G.J. Hamwi: "Familial gynecomastia." Ann. Int. Med., 63;377-385, 1965.

GRAVES-BASEDOW SYNDROME

Synonymy

S. de Von Basedow. S. de Flajani-Basedow. S. de Parry- - Serious. S. de Bergie. S. de Marsch. S. of hyperthyroidism. S. of thyrotoxicosis. S. of toxic goiter. S. of exophthalmic goiter.

Syndromography

Clínica

There is a female-male ratio of 4:1. It appears most frequently in the third to fifth decades of life. It is characterized by emotional disorders, nervousness, palpitations, heat intolerance, sweating especially in the hands; sometimes anorexia, nausea, vomiting, diarrhea and later dyspnea. Insomnia, weight loss with preservation of appetite.

Physical examination. Hot and moist skin, hyperpigmentation or vitiligo (associated), bilateral exophthalmos, rarely unilateral, infrequent blinking (Stellwag sign), inability to wrinkle the forehead when looking up (Joffroy sign), lazy eyelid (Von Graefe sign), loss of covergence (Rosenbach sign); diffuse, soft goiter, tachycardia, other arrhythmias (atrial fibrillation). The classic triad: diffuse goiter, exophthalmos, tachycardia. Hyperreflexia.

Paraclinical examinations

Electrocardiogramna. Tachycardia, arrhythmia.

Basal metabolism. High.

Blood. Increased protein-bound iodine (PBI), increased butanol-bound iodine (BBI), increased triiodotyranine (T3) and tetraiodotyranine (T4). Increased radioactive iodine uptake.

Syndromogenesis and etiology

Immunological; production of a slow-acting substance that stimulates the thyroid (thyrocytostimulating immunoglobulin) and another that produces exophthalmos (EF: exophthalmos factor).

Hyperthyroidism is the consequence of an excessive amount of hormone or an overactive hormone. The self-regulation mechanism of the pituitary-thyroid system is altered, but the cause that determines this is unknown. Psychological, stress, and somatic causes (traumas, infections, poisonings) influence. In any case there is an excess of iodine. Exophthalmos is due to the accumulation of mucopolysaccharides and water in the retrobulbar orbital tissue.

Literature

Flajani, G.: Osservazione LXVII: Sopra a freddo nellánterior tumor part of the dotto detto brancocele. In Collezione d'Osservazioni e Riflessioni di Chirurgia, G. Flajani. v. 3, S. Michele A. Ripa Presso Lino Contedini, Rome, 1802, p. 270.

Graves, R.J.: Clinical lectures. London, Med. Surg. J. (part II, 7;516, 1835.

Means, J.H.; L.J. De Groot and J.B. Stanbury: The thyroid and its diseases. ed. 3, New York. McGraw-Hill, 1963.

HANOT-CHAUFFARD SYNDROME

Synonymy

S. of diabetes-hemochromatosis. S. of tanned diabetes.

Iron deposition disease. Hemochromatosis. S. de Leschke. S. de Troisier-Hanot-Chauffard.

Syndromography

Clinic

This syndrome is much more common in men (80%) between 40 and 60 years old. There is lassitude, weakness, weight loss, skin pigmentation, acute pain in the right upper quadrant of the abdomen, dyspnea, loss of libido.

Physical examination. Skin pigmentation of two types: Addison-like or grayish tinge (genitals, face, arms, skin folds), rarely mucosal pigmentation (15%), edema, ascites. Late jaundice, body hair loss, gynecomastia, hepatomegaly, splenomegaly, testicular atrophy, eventual-mind heart failure.

Paraclinical examinations

Blood. At first there is a high concentration of hemoglobin, later macrocytic anemia, increased serum iron.

Hyperglycemia, hyperbilirubinemia. He-function tests altered pathology.

Liver biopsy.

Syndromogenesis and etiology

Inborn error of metabolism. Secondary forms due to iron overload.

The etiology is genetic with autosomal recessive inheritance.

Literature

Finch, C.A.: Hemochromatosis. In Beeson, P.B., and McDermott, W., ed:. : Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia, Saunders, 1967.

Hanot, V.C., et A.M. Chauffard: "Cirrhose hypertropique pigmentaire dans le diabetes sucré." Rev. of Med., 3:385-403, 1882.

Saddi, R. and J. Feingold: "Idiopathic haemochromatosis: an autosomal recessive disease." Clin. Genet., 5:234-241, 1974.

HARRIS SYNDROME

S. of organic hyperinsulinism. Neonatal hypoglycemia syndrome.

Syndromography

Clinic

The symptoms of this syndrome are the same as those of neonatal and chronic hypoglycemia, and sometimes there is irreversible brain damage.

It affects both sexes at all ages and occurs in the course of a wide variety of diseases, and as a presenting sign in many other syndromes.

It is characterized by Whipple's triad:

1. History of hunger attacks, weakness, sweating and facial paresthesias during fasting, tremor and palpitations. Incoherent speech, transient hemiplegia, seizures (cerebral response).

2. Low blood glucose during the crisis.

3. Immediate recovery with glucose administration.

Personality alterations: negativism, manic behavior (psychiatric response).

Central nervous system: diplopia, ataxia, aphasia, paralysis, seizures, coma.

Paraclinical examinations

Fasting for 72 hours (water only), moderate exercise. If the patient has an insular cell adenoma, blood glucose is reduced by 30%.

5-h glucose tolerance test. 1 g is administered intravenously, if there is an adenoma, after 30 minutes the blood glucose drops by 50-80% and remains low for several hours: if it is functional, the blood glucose falls but returns to normal in 1 or 2 hours.

Tolbutamide tolerance test. The excessive increase in insulin after tolbutamide is of great value, but more important is the prolonged persistence of hypoglycemia.

Leucine tolerance test.

Liver function tests.

Tolerance test to fructose-galactose, glucagon and epinephrine.

Blood. Very low blood glucose during crises. Insulin dosage. It should be remembered that hypoglycemic symptoms can appear with normal blood glucose and in some individuals with low blood glucose no symptoms appear.

Syndromogenesis and etiology

Multiple beta cell adenomas of the islets of Langerhans, small, rarely malignant, with functional metastasis. It is familial in nature and is frequently associated with adenomas of other endocrine glands (familial multiple adenomatosis).

The causes can be classified into two large groups:

Organic. Alteration of organs that intervene in the regulation of blood glucose. Extrapancreatic pancreatic tumors, liver and endocrine diseases, toxic and metabolic states.

Functional. When no organic cause for hypoglycemia can be established.

Literature

Harris, S.: "Hyperinsulinism and dysinsulinism." J.A.M.A., 83:729-733, 1924.

TRANSIENT HYPOGLYCEMIC HEMIPPLEGY SYNDROME

Syndromography

Clínic

It affects both sexes at any age. Seizures can be precipitated by food or exercise. Transient flaccid or spastic paralysis is observed, with a variable degree of muscle weakness that most frequently involves the right side.

It can end in a few hours or days, frequently recurring about three times in a few days.

Physical examination. Babinski positive, often bilateral.

Inconstant changes of the sensorium. Tachycardia, profuse sweating preceding paralysis and convulsions (frequently coma). Occasionally, however, hemiplegia may be an isolated manifestation.

Paraclinical examinations

Blood. Hypoglycemia (investigate cause).

Syndromogenesis and etiology

Hypoglycemia of any origin (endogenous or exogenous): insulin administration, adrenal tumor, pancreatic adenoma, idiopathic.

Literature

Miller, W.L. and J.H. Trescher: “Amnesia, epileptiform convulsive seizures and hemiparesis as manifestations of insulin shock." Amer. J. Med. Sci., 174:453, 1927.

Montgomery, B.M. and C.A. Pinner: "Transient hypoglycemic hemiplegia." Arch. Int. Med. 114, 680-684, 1964.

HERS SYNDROME

Synonymy

S. of hepatic phosphorylase deficiency. S. of glycogenosis due to hepatophosphorylase deficiency. Cori type VI glycogenosis syndrome.

Syndromography

Clinic

It affects both sexes and begins in childhood. The symptoms may be so moderate that the syndrome may pass

unnoticed. There is growth retardation and Harris syndrome (see this one). Obesity of the trunk with characteristic "bambola" facies. Tendency to vomit with acidosis crisis

Physical examination. Great hepatomegaly.

Paraclinical examinations

Blood. There is deficiency of phosphorylase activity in leukocytes. Normal glycogen in erythrocytes and leukocytes.
During prolonged fasting: hypoglycemia and ketosis.
Hyperlipidemia and sometimes hyperbilirubinemia and abnormal liver tests

Syndromogenesis and etiology

In these patients there is a reduction in hepatic and leukocyte glycogen-phosphorylase, greater than 25-50% of normal. The relationship between the biochemical defect and the clinical manifestations is the consequence of a defect in the breakdown of glycogen.

The etiology is genetic due to autosomal recessive inheritance.
The gene for the enzyme hepatic glycogenophosphorylase has been assigned to chromosome 14.

Literature

Field, R.A.: Glycogen deposition diseases. In Stanbury, J.B., J.B. Wyngaarden, and D.S. Frederickson; The metabolic basis of inherited disease. ed. 2, New York, McGraw-Hill, 1966.
Hers, H.G.: "Etudes enzymatiques sur fragments hepatiques application a la classification des glycogenoses." Rev. Intern. d'Hepatol., 9:35-55, 1959.
Newgard, C.B. et al.: "The polymorphic locus for glycogen storage disease VI (liver glycogen phosphorylase) map to chromosome 14". Am. J. Hum. Genet., 40:351-364, 1987.

HPERBETALIPOPROTEINEMIA SYNDROME

Synonymy

S. of familial hyperlipoproteinemia type II. S. of familial hypercholesterolemia. S. of essential familial hypercholesterolemia. Familial hypercholesterolemia-xanthomatosis syndrome. S. of familial xanthoma. Simple tuberous xanthoma S.

Syndromography

Clinic

Affects both sexXOS; It is detected from childhood to the third or fourth decade of life; anginal pain and symptoms of arterial obstruction in different organs.

Physical examination. Tendonous or tuberous xanthoma especially on the elbows, knees, hands and feet; senile arch; not hepatosplenomegaly.

Paraclinical examinations

Blood. Increase in betalipoprotein. Increase in cholesterol more than phospholipids. Normal triglyceride test. Normal glucose tolerance test.

Electrocardiogram.

Radiology. Calcification of vascular atheromas.

Syndromogenesis and etiology

Inherited defect in the control of plasma lipoproteins.

A gene abnormality is proposed that interferes with the regulation of the activity of the 3-hydroxy 3-methylglutaryl coenzyme A reductase, which controls the rate of cholesterol synthesis.

There is genetic heterogeneity, most mutations segregate as an autosomal dominant inheritance.

Literature

Rayer, P.E.O.: Traite theorique et practiques des maladies de la peau. Paris, 1836.
Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The metabolic basis of inherited disease. ed., 2, New York, McGraw-Hill, 1966.

FAMILIAR HYPERPREBETALIPOPROTEINEMIA SYNDROME

Synonymy

S. of familial hyperlipoproteinemia type IV. S. of carbohydrate-induced hyperlipidemia. S. of essential familial hyperlipidemia.

Syndromography

Clinic

It affects both sexes and is detected between the second and sixth decades of life. Uncertain clinical findings. Ischemic heart disease.

Physical examination. Eruptive xanthomas may or may not be present.

Paraclinical examinations

Blood. Elevation of prebetalipoprotein independent of diet; constant elevation of triglycerides (with wide variation), cholesterol and generally normal phospholipids. Abnormal glucose tolerance test, occasional hyperglycemia.

Electrocardiogram.

Syndromogenesis and etiology

Family inheritance of undetermined type. Endogenous form of hyperlipoproteinemia not related to diet. It is related to ischemic heart disease, although less closely than type I.

The phenotype is strongly influenced by environmental factors, particularly by carbohydrate and ethanol consumption.

Family reports suggest an autosomal dominant inheritance.

Literature

Stanbury, J.B.: J.B. Wyngaartden and D.S. Frederickson: The metabolic basis of inherited disease. ed. 2. New York, McGraw-Hill, 1966.

DEHYPERCALCEMIA SYNDROME

Syndromography

Clinic

Malaise, muscle weakness, polyuria, polydipsia, constipation, dehydration, nausea, vomiting, anorexia.

Mental disorders from depression to delirium.

Physical examination. Cardiac arrhythmias, muscle hypotonia, areflexia.

Paraclinical examinations

Blood. Elevation of calcium levels, associated with other typical findings of various pathological entities.

Electrocardiogram.

Syndromogenesis and etiology

Hypercalcemia can be seen in several different conditions: bone cancer, myeloma, sarcoidosis, hypervitaminosis D, primary and secondary hyperparathyroidism, Burnett syndrome, hypophosphatasia syndrome, prolonged immobilization, treatment with estrogens or testosterone.

Literature

Goldsmith, R.S. and s.h. Ingbar: "Inorganic phosphate treatment of hypercalcemia of diverse etiologies." N. Engl. J. Med., 274:1-7, 1966.

HYPERKALIEMIC SYNDROME

Synonymy

S. of potassium poisoning.

Syndromography

Clínic

This syndrome is asymptomatic in the moderate form. In the severe form there is muscle weakness and flaccid paralysis.

Physical examination. The early signs are provided by electrocardiographic changes with high, peaked T waves and decreased P waves.

Paraclinical examinations

Blood. Elevated serum potassium.

Urine. Elevated urinary potassium.

Electrocardiogram. Signs of hyperkalemia.

Syndromogenesis and etiology

Renal failure, diabetic ketoacidosis, post-surgical electrolyte disorders, Addison syndrome, Conn syndrome. Iatrogenesis due to spironolactone, trianterene, blood transfusion or excessive administration of potassium.

Literature

Schwartz, W.B.: Fluid, electrolyte, and acid-base balance. In Beeson, P.B. , and W. McDermot, ed.: Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia, Saunders, 1967.

HYPERSTIMULATION SYNDROME

Synonymy

Meig's syndrome secondary to ovarian stimulation.

Syndromography

Clínic

Patients receiving human gonadotropin for the treatment of amenorrhea and infertility. Abdominal pain and distension and acute abdominal crises, tachycardia, oliguria, hematuria, chest pain, dyspnea.

Physical examination. Ascites, hydrothorax, large ovaries.

Paraclinical examinations

Ionogram.

Electrocardiogram.

Chest radiology.

gynecological ultrasound.

Syndromogenesis and etiology

Administration of human gonadotropin, clomiphene.

Literature

Neuwirth, R.S.; R.N. Tuksoy and R.L. Vande Wiele: *Acute Meig's syndrome secondary to ovarian stimulation with human menopausal gonadotropins". Amer. J. Obstet. Gynecol., 91:977- -981, 1965.

Vandiest, L. and DeBast, A.: “Syndrome abdominal aigu par dégénérescence kystique massive et totale des ovaries due to administration des gonadotrophines.” Bruxelles Med., 38:1636- -1642, 1958.

ANTERIOR PICTURE HYPERFUNCTION SYNDROME

Syndromography

Clinic

It is a rare syndrome. It occurs in patients between 20-30 years old. There are manifestations of hyperpolymenorrhea, anemia, galactorrhea, hyperthyroidism (tachycardia, sweating, diarrhea), nervousness, weight loss, secondary sterility.

Physical examination. Pallor, emaciation, goiter with hyperthyroidism (exophthalmos), flaccid breasts with areolar hyperpigmentation, enlarged uterus, palpable

Paraclinical examinations

Blood. Anemia, hypoglycemia and occasionally a flat curve in the glucose tolerance test.

Urine. Increase in estrogen without increase in gonadotropins (FSH).

Vaginal smear. Hyperstrinism.

Hyterosalpingography. Normal.

Milk. Does not coagulate when boiling; elevated prolactins, increased TSH; T3-T4 increased.

Radiology. Normal Turkish chair.

Syndromogenesis and etiology

Overproduction of estrogens, thyrotropin, prolactin, due to anteropituitary hyperplasia or hypothalamic dysfunction.

Literature

Dowling, J.T.; J.B. Richards; N. Freinkel, et al.: "Nonpuerperal galactorhea." Arch. Int. Med., 107:885-893, 1961.

Zondek, B.; Y.M. Bromberg and S. Rozin: “Anterior pituitary hyperchromonotrophic syndrome (excessive uterine bleeding, galactorrhea, hyperthyroidism).” J. Obstet. Gynaec. Brit. Emp., 58:525-537, 1951.

FAMILIAR BETAPREBETA HYPERLIPOPROTEINEMIA SYNDROME

Synonymy

S. of familial hyperlipoproteinemia type III. S. of familial hypercholesterolemia with hyperlipidemia. S. of hyperlipidemia in familial hypercholesterolemic xanthomatosis. S. of carbohydrate-induced hyperlipidemia. S. of essential familial hyperlipidemia.

Syndromography

Clinic

It affects both sexes and is detected in the third to fourth decades of life. It is characterized by anginal pain and symptoms of arterial obstruction in different organs. Abdominal pain crises may occur.

Physical examination. Appearance of nodules in tendons in various locations and other flat or tuberous xanthomas. Hepatosplenomegaly.

Paraclinical examinations

Blood. Hyperlipidemia, hypercholesterolemia, decreased prebetalipoprotein. Abnormal glucose tolerance test, sometimes frank diabetes. Hyperuricemia.

Electrocardiogram.

Radiology. It allows demonstrating calcification of atheromas.

Syndromogenesis and etiology

The genetic etiology of this syndrome is related to the characteristics of the apolipoprotein E (apoE) genes, which is a very low-density lipoprotein (VLDL). The existence of a simple locus with three common alleles that produces alpha II, alpha II and alpha IV and beta II, beta III and beta IV patterns in genotypic combinations has been proposed. Beta IV subclass is the one associated with type III hyperlipoproteinemia.

In normal individuals, chylomicrons and remaining VLDL are rapidly removed from the circulation by receptor-mediated endocytosis in the liver.

In families, this type III of hyperlipoproteinemia, the increase in plasma and triglycerides, is the consequence of abnormality in the plasma clearance of chylomicrons and remaining VLDL due to the apoE defect.

This produces xanthomatosis and coronary and/or peripheral vascular disease.

Type III hyperlipoproteinemia may be due to a primary hereditary defect (homozygous people) for the E2 isoform, but only 1-4% of E2/E2 homozygotes develop this defect, which is why the presence of secondary factors such as hypothyroidism, systemic lupus erythematosus or diabetic acidosis has been proposed that may precipitate this defect in heterozygous individuals.

The phenotype depends on age and is rarely observed in the third decade of life.

Molecular investigations have shown that the apoE gene is located on chromosome 19g.

Literature

Malmros, H.; B. Swahn and E. Truedsson: "Essential byperlipemia." Acta Med. Scand., 149:91, 1954.

Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The metabolic basis of inherited disease. ed.2, New York, McGraw-Hill, 1966.

Zannis, V.I.; P.W. Just and J.L. Breslow: Human apolipoprotein E isoprotein subclasses and genetically determined". Am. J. Hum. Genet., 33:11-24. 1981.

MIXED HYPERLIPOPROTEINEMIA SYNDROME

Synonymy

S. of familial hyperlipoproteinemia type V. S. of hyperchylomicronemia and familial hyperprebetalipoproteinemia. S. of hyperlipoproteinemia induced by excess fat and carbohydrate. S. of essential familial hyperlipoproteinemia.

Syndromography

Clinic

It affects adults of both sexes. It is manifested by recurrent abdominal pain in relation to the ingestion of a large amount of fat, occasionally mild diabetes. Rarely eruptive xanthomas. No other manifestations.

Paraclinical examinations

Blood. Increase in chylomicrons and prebetalipoprotein unrelated to diet. Abnormal glucose tolerance test.

Lipidogram. VLDL increased, LDL and HDL decreased on an empty stomach.

Syndromogenesis and etiology

There are several conditions that cause this phenotype: insulin-dependent diabetes mellitus, the use of steroid contraceptives, alcohol abuse, and glycogen storage disease type I.

From the genetic point of view, only one three-generation family has been reported, but the mode of inheritance has not been defined.

Literature

Malmros, H.; B. Swahn, and E. Truedsson: "Essential hyperlipemia". Acta Med. Scand., 149:91-108, 1954.

Francois, J, et al.: "Genetic study of hyperlipoproteinemia types IV and y." 12:202-207, 1977.

HYPERLYSINEMIA SYNDROME

Syndromography

Clinic

This syndrome presents from birth, with seizures, insufficient development, and severe mental retardation.

Physical examination. Flaccidity of muscles and ligaments, stopping river of sexual development.

Paraclinical examinations

Permanent increase in lysine in blood and urine that contrasts with the temporary increase observed in other syndromes. Homocitrulline and homoarginine are also present in urine.

Syndromogenesis and etiology

It is due to a defect in the enzyme aminoradipien semialdehyde synthetase (AASS), which is a hometetramer with

enzymatic activity for the degradation of lysine (lysine ketoglutarate reductase) and saccharopine.

It is transmitted by autosomal recessive inheritance.

Literature

Colombo, J.P.; W. Burgi; R. Richterich, et al.: "Congenital lysine intolerance with periodic ammonia intoxication: a defect in L-lysine degradation." Metabolism, 16:910-925, 1967.

Cox, R.P. et al.: "Multiple enzyme deficiency in familial hyperlysinemia." (Abstract). Am. J. Hum. Genet., 27:59A, 1975.

Ghadimi,H.; V.I. Bimington and F. Pecora: "Hyperlysinemia associated with retardation." N. Engl. J. Med., 273:723-729, 1965.

HYPERMAGNESEMIA SYNDROME

Syndromography

Clinic

Nausea, vomiting, malaise, urinary incontinence, depression of the central nervous system (drowsiness, lethargy, slight slurred speech), ataxia. Eat (when there are very high concentrations). Cardiac arrest.

Physical examination. Arterial hypotension with high concentrations, osteotendinous areflexia,

Paraclinical examinations

Blood. Hypermagnesemia.

Electrocardiogram. Prolonged P-R, slight decrease in the Py wave of the QRS duration, nodal rhythm with bundle branch block, increased QT.

Syndromogenesis and etiology

In newborns of mothers who have received large doses of magnesium intravenously during prepartum. In individuals who ingest large doses of magnesium, antacids or receive magnesium salts through this route and in patients with renal failure who have great difficulties excreting magnesium.

Literature

Lipsitz, P.J. and I.C. English: "Hypermagnesemia in the newborn infant." Pediatrics, 40:856-862, 1967.

Randall, R.E.; M.D. Cohen; DC Spary, et al.: "Hypermagnesemia in renal failure; etiology and toxic manifestations." Ann. Int. Med., 61:73-88, 1964.

PRIMARY HYPEROXALURIA SYNDROME

Synonymy

S. of hyperoxaluria. S. of oxalosis. S. de Lepoutre.

Syndromography

Clinic

It occurs in adults at an early age. Nephrolithiasis and nephrocalcinosis progressing to chronic renal failure.
Some congenital cases begin in childhood and are manifested by nausea, vomiting, dry and toasted mouth, abdominal pain, renal colic, and occasional tetany.

Paraclinical examinations

Blood. Increased oxalates, low calcium. Bone marrow: calcium oxalate crystals.

Urine. Excretion of oxalates, 3-5 times above normal, albuminuria, hematuria, calcium oxalate casts and crystals.

Radiology. Osteoporosis, bilateral kidney stones.

Syndromogenesis and etiology

In the congenital form, autosomal recessive inheritance is mentioned. In the acquired form, metabolic disorders with the accumulation of oxalic acid in the tissues.

Type I. Glycolic aciduria, 2-0xoglutarate-glyoxylate carboligase defect.

Type I. L-glyceric aciduria, D-glyceric dehydrogenase defect.

There are many other acquired forms where calcium oxalate accumulates in the tissues: oxalate poisoning, ethylene glycol poisoning, glyocoxylate administration, pyridoxine deficiency, liver cirrhosis, renal tubular acidosis syndrome.

This defect is due to mutations in the gene that codes for the liver enzyme peroxisomalalanine glycoxylate aminotransferase.

In some patients it is reported that the reduction in enzymatic activity is due to the absence of peroxisomes.

In 1992, McKusick codified four types of primary hyperoxaluria type I due to different types of mutations of the AGXT gene (symbology used for this gene). At the same time, the AGXT gene has been located on chromosome 2q36-q37.

Literature

Lepoutre, C.: "Calculs multiples chez un enfant: Infiltration du parenchyme renal par des depots cristallins." J. Urol. Paris, 20:424, 1925.

Williams, H.E. and L.H. Smith, Jr.: "1-Glyceric aciduria, a new genetic variant of primary hyperoxaluria." N. Engl. J. Med., 278:233-239. 1968.

Purdue, P.E. et al.:"Characterization and chromosomal mapping of a genomic clone encoding human alanine: glyoxylate aminotransferase." Genomics, 10: 34-42, 1991.

FAMILIAR HYPERPARATHYROIDISM SYNDROME

Syndromography

Clínic

Variable manifestations of hyperparathyroidism in members of the same family. Frequent association with glandular pathology. More frequent association with peptic ulcer, pancreatitis and kidney stones (see polyendocrine syndrome).

Paraclinical examinations

Blood. Calcium, phosphorus, alkaline phosphatase.

Kidney ultrasound.

Radiology. Descending urogram, gastrointestinal tract.

Syndromogenesis and etiology

It is a syndrome of genetic etiology with autosomal dominant inheritance. The basic defect is unknown.

Literature

Cutler, R.E.; E. Reiss; L.V. Ackerman: "Familial hyperparathyroidism. A kindred involving eleven cases with a

discussion of primary chief-cell hyperplasia", N. Engl. J. Med., 270:859-865, 1964.

Goldman, L. and F.S. Smith: "Hyperparathyroidism in siblings." Ann. Surg., 104:971-981, 1936.

SEVERE SECONDARY HYPERPARATHYROIDISM SYNDROME

Synonymy

S. of renal osteodystrophy.

Syndromography

Clínic

Advanced chronic renal failure as the basis of the process. Bone pain, ectopic calcification and pruritus. Osteomalacia, cystic fibrosa osteitis.

Paraclinical examinations

Kidney function should be studied to detect advanced chronic kidney failure if this is the cause.

Blood. Hypocalcemia, increased paratormone.

Syndromogenesis and etiology

Parathyroid hyperplasia, with the resulting increase in parathyroid secretion, is due to resistance to the normal concentration of the hormone, which causes hypocalcemia, which in turn stimulates parathyroid growth and occurs in patients with renal failure, osteomalacia (vitamin D deficiency) and pseudohypoparathyroidism (deficient response of parathyroid receptors).

OVARIAN HYPERTHECOSIS SYNDROME

Synonymy

S. of diffuse luteinization of the ovarian stroma.

Syndromography

Clinic

Amenorrhea, sterility.

Physical examination. Hirsutism, occasionally virilism.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Gold, J.J.: Textbook of gynecologic endocrinology. New York, Harper & Row, 1968. p.320.

HYPERTONICITY SYNDROME DUE TO SOLUTE OVERLOAD

Synonymy

S. of water loss-excess solutes.

Syndromography

Clinic

The same as pure water loss syndrome with two exceptions: the patient gains weight instead of losing it and is polyuric with hyposthenuria instead of oliguric.

Paraclinical examinations

Urine. Polyuria with low density.

Syndromogenesis and etiology

Nasogastric tube feeding without adequate water intake. Digestive bleeding treated with milk and alkali, without water adequately. Children fed only highly concentrated milk. Diabetics with polyuria treated with excessive intake of electrolytes and insulin.

Literature

Goldberger, E.: A Primer of water, electrolyte, and acid-base syndromes. ed. 3, Philadelphia, Lea & Febiger, 1965.

HYPERVALINEMIA SYNDROME

Synonymy

S. of valinemia.

Syndromography

Clinic

It occurs from early childhood, there is vomiting, diarrhea, insufficient development, mental retardation, hyperkinesia.

Physical examination. Muscle hypotonia, nystagmus.

Paraclinical examinations

Blood and urine. Increased valine concentration.

Electroencephalogram.

Electromyogram.

Syndromogenesis and etiology

Deficiency of the enzyme that converts valine into its corresponding keto acid.

It is a defect of genetic etiology and autosomal recessive inheritance. The basic defect is represented by deficiency of the enzyme valine transaminase.

Literature

Budd, M.A.: K. Tanaka; L.B. Holmes, et al.: Isovaleric acidemia". N. Engl. J. Med., 277:321-327, 1967.

Dancis, J. et al.: "Hypervalinemia: a defect in valine transamination." Pediatrics, 39:813-817, 1967.

Wada, Y., et al.: "Idiopathic hypervalinemia: probably new entity of uniform error of valine metabolism." Tohuku J. Exper. Med., 81:46-55, 1963.

HPOCALCACEMIC SYNDROME

Syndromography

Clinic

Tetany. In children: laryngospasm, facial muscle spasms, paresthesias, character changes, asthmatic attacks, convulsions.

Physical examination. Trousseau and Chvostek signs; dry and thick skin, sparse or absent hair, sparse hair. Other signs in relation to the underlying disease.

Paraclinical examinations

Blood. Hypocalcemia, variable potassium.

Urine. Absence of calcium, Bllsworth-Howard test.

Electrocardiogram. Prolonged Q-T, normal S-Ty T.

Stool.

Radiology. According to the etiology.

Syndromogenesis and etiology

1. Rickets and osteomalacia.
2. Intestinal malabsorption.
3. Fitz syndrome.
4. Hypoparathyroidism.
5. Pseudohypoparathyroidism.
6. Kidney failure.
7. Epstein syndrome.
8. Use of chelating agents.
9. Use of citrated blood in large quantities.

HPOCALIEMIC SYNDROME

Synonymy

Hypokalemic S. (See also familial Mediterranean fever syndrome.)

Syndromography

Clinic

Anorexia, nausea, vomiting, abdominal distension, paralytic ileus, weakness, sensory depression. Occasional thirst and polyuria. Tetany may occur (see alkalosis syndrome).

Physical examination. Abdominal tympany, tendon hyporeflexia.

Paraclinical examinations

Blood. Low potassium, normal, low or high calcium, evidence of metabolic alkalosis.

Urine. Variable potassium.

Electrocardiogram. (Important exam.) T flattened and wide, Q-T prolonged and if hypokalemia worsens the T becomes negative, the S-T is depressed and a U wave appears.

Syndromogenesis and etiology

1. Excessive administration of fluids.
2. As part of alkalosis syndrome.
3. Loss of potassium through urine and feces.
4. Chronic malnutrition.
5. Redistribution of potassium in the body (muscle, liver).

Literature

Goldberger, E.: A primer of water, electrolyte, and acid-base syndrome. ed. 3, Philadelphia, Lea & Febiger, 1965.

HYPOPHOSEATASIA SYNDROME

Synonymy

S. of Rathbun.

Syndromography

Clinic

It predominates in women and varies according to the age of onset. In the fetus there is a deficiency of the bone support of the skull and thorax. In childhood, nausea and vomiting (see hypercalcemic syndrome), rickets-like changes, bowing of the legs, prominent chondrocostal joints, genu valgus, exophthalmos, symptoms and signs of intracranial hypertension, seizures. Premature loss of teeth.

In adults there are asymptomatic or occasional fractures. Blue sclerae.

Paraclinical examinations

Blood. Low alkaline phosphatase, hypercalcemia.

Urine. Presence of phosphoethanolamine, hypercalciuria.

Bone biopsy. Typical histological changes.

Radiology. Nicked epiphyses; skull with large areas of osteolysis and premature calcifications; the sutures show grooved bony edges. Pseudofractures.

Syndromogenesis and etiology

Autosomal recessive hereditary transmission. There is also a dominant form. Heterozygotes are recognized by hypophosphatasia and phosphoethanolaminuria.

The etiology is unknown.

Literature

Bartwell, W.M. Jr.; L. Riggs; L.F.A. Peterson, et al.: "Hypophosphatasia in an adult." Arch. Int. Med., 120:90-93, 1967.

Huhne, T. and E. Schonfeld: Eine Eigenartige Wachstrumstorung im Kindesalter". Monatsschr. Kinderch., 42:267, 1929.

HPOMAGNESEMIA SYNDROME

Synonymy

S. of magnesium deficiency.

Syndromography

Clinic

.Physical examination. Signs of Chvostek, negative Trousseau.

Paraclinical examinations

Determination of magnesium, calcium and blood proteins.

Syndromogenesis and etiology

Magnesium deficiency due to gastroenteritis, malnutrition and diarrhea. Increase in calcium intake after profuse diarrhea. A possible primary hereditary disorder is also mentioned.

Literature

Flink, E.B.; R. McCollister; ACE. Prasad, et al.: "Evidences for clinical magnesium deficiency." Ann. Int. Med., 47:956-968, 1957.

Friedman, M.; G. Hatcher and L. Wats on: *Primary hypomagnesaemia with secondary hypocalcemia in an infant". Lancet, 1:703-705, 1967.

Savage, D.C.L. and W.A.F. McAdam: "Seizures due to hypomagnesaemia in an infant recovering from diarrhea."
Lancet, 2:235-236, 1967.

HYPONATREMIA SYNDROME

Synonymy

S. of sodium loss. S. of pure salt depletion. S. of hyposmolarity. S. of hypotonic dehydration. S. de Schroeder I.

Syndromography

Clínica

In acute loss there is shock; in the chronic, weakness, apathy, lassitude, absence of thirst, anorexia, nausea, muscle cramps, orthostatic hypotension, cold limbs, progression to confusion and acute cerebral syndrome (see this one). Oliguria and finally kidney failure.

Physical examination. Sunken, glassy eyes; wrinkled tongue, skin fold, tachycardia, arterial hypotension, weak pulse, venous collapse. In severe forms, the skin is sticky and sweaty.

Paraclinical examinations

Blood. Hemoconcentration, with increased mean corpuscular volume of erythrocytes. Hypovolemia. Low sodium, chlorine and bicarbonate, usually high potassium. Elevated urea nitrogen. Low osmolarity.

Urine. Low volume, normal density, very low sodium and chlorine.

Syndromogenesis and etiology

Sodium loss: vomiting, diarrhea, gastrointestinal or biliary fistula, gastric catheterization and aspirations, profuse sweating (with ingestion of water), burns, as part of mucoviscidosis.

2. Paralytic or mechanical ileus, serous loss.
3. In the immediate postoperative period, in Addison's syndrome, Thorn syndrome, inadequate secretion of antidiuretic hormone, diabetic.

4. Iatrogenic: administration of diuretics or forced diuresis, in secondary aldosteronism and sodium retention syndromes (see Schroeder syndrome), Hypodermoclysis of dextrose in water.

Literature

Bartter, F.C.: "Hyper and hypo-osmolality syndromes." Amer. J. Cardiol., 12:650-655, 1963.
Goldberger, E.: A primer of water, electrolyte, and acid-base syndromes. ed. 3, Philadelphia, Lea & Febiger, 1965.
Schroeder, H.A.: "Renal failure associated with low extracellular sodium chloride, the low salt syndrome." J.A.M.A., 141:117- -124, 1949.

CHRONIC DILUTIONAL HYPONATREMIA SYNDROME

Synonymy

S. of chronic cellular hyposmolarity. S. of internal dilutional hyponatremia. S. of asymptomatic hyponatremia. S.de essential hyponatremia. S. of new permanent regime. S. of the sodium paradox.

Syndromography

Clinic

It is observed in patients with advanced congestive heart failure (treated or not), prolonged wasting diseases, malnutrition, severe chronic infection. It does not have specific symptoms and basically the laboratory is what shows hyponatremia, or the symptoms and signs are those of Schroeder syndrome.

Physical examination. Lethargy, anorexia, disorientation, edema, ascites, pleural effusion.

Paraclinical examinations

Blood. Low sodium and potassium. Anemia with decreased corpuscular volume. Normal creatinine. Hypoproteinemia, estimation of total body water and electrolyte values reveal normal sodium values.

Urine. Elevated potassium may be found.

Syndromogenesis and etiology

Exchange of sodium ions for potassium that is continuously released from the cells, two sodium ions replace three potassium ions with a drop in cellular osmotic pressure, consequent secretion of ADH and water retention, which causes a decrease in the concentration of extracellular sodium. It should not be confused with Schroeder syndrome (see this one) which occurs in intensively treated congestive heart failure and presents the same finding of hyponatremia.

Literature

LaRotonda, M.L. and W.J. Grace: The low salt syndrome. an extreme example in heart failure". Amer. J. Cardiol., 6:676- -677, 1960.
Vogl, A.: "The low-salt syndrome in congestive heart failure." Amer. J. Cardiol.. 3:192-198. 1959.

HYPOPARATHYRODEUS SYNDROME

Syndromography

Clinic

It starts at any age. It is characterized by muscle spasms and tetany, nervousness, weakness, paresthesias in the hands, blurred vision, headache, memory loss.

Physical examination. Chvostek and Trousseau signs.
Papilledema, nail and tooth malformations, thick and dry skin, alopecia, cataracts.

Paraclinical examinations

Blood. Hypocalcemia, hyperphosphoremia.
Radiology. Increased bone density, basal ganglia calcification.

Electrocardiogram.
The therapeutic trial with paratormone produces acute phosphaturia.

Syndromogenesis and etiology

This syndrome occurs in:

1. Families with the isolated autosomal recessive juvenile type (without Addisonian component).

2. Idiopathic acquired juvenile.
3. Idiopathic or acquired after thyroid surgery.

Literature

Deiss, W.P., Jr.: “Diseases of the parathyroid glands.” Med Times, 95:633-647. 1967.

CONSTITUTIONAL HIRSUTISM SYNDROME

Synonymy

S. of idiopathic hirsutism.

Syndromography

Clinic

Affects the female sex.

Physical examination. Increase in body hair (face, chest, abdomen and limbs), without changes in secondary sexual characteristics.

Paraclinical examinations

Blood. Normal hormonal secretion and excretion. See other hirsutisms.

Syndromogenesis and etiology

Familiar? Racial?
The etiology is unknown.
Women carriers of mutations that generate 21-hydroxylase deficiency usually present hirsutism, so this entity must be kept in mind.

HISTIDINEMIA SYNDROME

Syndromography

Clinic

It begins before the age of 8 and is sometimes discovered in neonatal amino acid testing.
In half of the cases it is asymptomatic. There is moderate to severe mental and physical retardation, speech disorders with difficulty in pronouncing consonants. Sometimes cerebellar ataxia, epilepsy, hydrocephalus, obesity, precocious puberty, bone defects, light eyes and hair, Marfan syndrome.

Paraclinical examinations

Blood. Histidine elevated 4-10 times normal (1-1.5 mg, 100 mL).

Urine. Histidine increased 6-10 times normal (30-60 mg in 24 hours). Positive ferric chloride test.
Cerebrospinal fluid. Normal or elevated histidine.

Syndromogenesis and etiology

Genetics: autosomal recessive, some dominant cases. One case every 16,000 births. The absence of hepatic histidine forces us to use another route for the metabolism of histidine, converting it into urocanic acid and finally pyruvic acid that appear in the urine. In half of the cases there is brain damage in the first two years.

Literature

Ghadim, H. and M.W. Partington: "Salient features of histidinaemia." Am. J. Dis. Child., 113: 83. 1967.

Griffiths, M.: Clinical aspects of the dietary treatment of histidinaemia in: Seakins and Saunders (Eds): Treatment of Inborn Errors of Metabolism. Ed. Churchill Livingstone, Edinburgh, 1973, p. 87.

HOFFMAN II SYNDROME

Synonymy

S. of myopathy-myxedema. S. of myotonic dystrophy and myxedema.

This eponym is used to designate the muscular manifestations described in Kocher-Debré-Semelaigne syndrome associated with myxedema in children and adults.
There is probably no fundamental difference between Hoffman syndrome and Kocher-Debré-Semelaigne syndrome.

Literature

Adams, R.D. In Werner, S.C.. ed.: The Thyroid, ed. 2, New York.
Hoeber Harper Row, 1962.

Hoffman, J.: Weiterer Beitrag zur Lehre von der Tetanie. Deutsch.
Ztschr. Nervenh.. 9:278-290, 1897.

HOUSSAY SYNDROME

Synonymy

S. of evanescent diabetes mellitus.

Syndromography

Clinic

The symptoms and signs of diabetes disappear while those of hypopituitarism become suddenly or progressively evident. There is usually little or no evidence of specific clinical events except in the fulminant type of hypopituitarism.

Paraclinical examinations

Blood. Hypoglycemia, persistence of tolerance curve
abnormal.

Urine. Decrease in gonadotropins, corticosteroids and 17-ketosteroids.

Syndromogenesis and etiology

Necrosis or pituitary infarction in diabetics.

Literature

Calvert, R.J. and G Caplin: "The Houssay syndrome." Br. Med.
J.. 2:71-74, 1957.

Houssay, B.A. and A. Biasotii: "Pancreatic diabetes of hypophysis-deprived dogs." Rev. Soc. Argent. de Biol., 6:251-296, 1930.

CORPUS LLUIEUS INSUFFICIENCY SYNDROME

Syndromography

Clinic

History of abortions, particularly frequent abortions.

Paraclinical examinations

Blood. Low levels of pregnandiol excretion during the initial phase of the menstrual cycle.

Endomnetrium biopsy (day 24 of the cycle) poor secretion, and low storage of glucose, alkaline phosphatase and other enzymes.

Syndromogenesis and etiology

It is due to poor endogenous secretion of progesterone during the luteal phase of the menstrual cycle.

Literature

Kupperman, H.S.: "Treatment of endocrine causes of sterility in the female." Clin. Obstet. Gynec., 2:808-825, 1959.

METABOLIC INSUFFICIENCY SYNDROME

Synonymy

S. of hypometabolic state.

Syndromography

Clinic

It can be asymptomatic or with vague and non-specific symptoms: fatigue, obesity, menstrual irregularity, dry skin and hair, emotional disorders.

Paraclinical examinations

Thyroid function tests. Normal with low basal metabolism.

Syndromogenesis and etiology

This syndrome is not very well defined and there is not enough information to guarantee its acceptance.

Literature

Morton, JH.: "Sodium liothyronine in metabolic insufficiency syndrome and associated disorders." J.A.M.A., 165:124-129, 1957.
Paschkis, K.E.; A.E. Rakoff; A. Cantarow, et al.: Clinical Endocrinology. ed. 3, New York, Harper-Row, 1967.
Thurmon, F.M. and W.O. Thompson: "Low basal metabolism without myxedema." Arch. Int. Med., 46:879-897, 1930.

ACUTE ADRENAL INSUFFICIENCY SYNDROME

Synonymy

S. of addisonian crisis. S. de Bernard-Sergent.

Syndromography

Clínica

It begins dramatically: anorexia, nausea, vomiting, abdominal pain, cyanosis, mental dullness.

Physical examination. Weak pulse, severe hypotension, weak heart sounds, dehydration, hypothermia. Fever appears when treatment begins.

Paraclinical examinations

Do not delay rapid treatment waiting for tests. Blood glucose, sodium, potassium, blood count, plasma cortisol.

Syndromogenesis and etiology

Adrenal insufficiency due to bilateral hemorrhage; in the course of infections, especially meningococcus (very serious and almost always fatal, known as Waterhouse-Friderichsen syndrome), trauma, tumor, thrombosis;
bilateral adrenalectomy without prior replacement treatment;
cortical atrophy and sudden withdrawal of cortisone treatment,
stress of a different nature with a decrease or inability to increase hormone production.

Bibliography

Addison, T.: Diseases of adrenal capsules. London, Med. Gaz. YOU. 43(N.S. 8):517-518, 1849.
Farwleg, F.F.: Adrenal cortical insufficiency. In Eisentein, A.B. (ed.). The adrenal Corted, Boston, Little Brown, 1967.
Friederichsen, C.: "Nebennierenapoplexie bei kleinen Kinder." Jb. Kinderhk. 87:109-125, 1918.
Turkington, R.W. and H.E. Lebovitz: "Extrarenal endocrine deficiencies in Addison's disease." Amer. J. Med., 43:499-507, 1967.

Waterhouse, R.: *A case of adrenal apoplexia". Lancet II:577-578. London 1911.

CHRONIC ADRENAL INSUFFICIENCY SYNDROME

Synonymy

Addison's disease.

Syndromography

Clinic

Early emotional lability, irritability. Appearance of the following symptoms: asthenia, weakness, facial fatigue (improves with rest), anorexia, nausea, vomiting, weight loss, abdominal pain, constipation or diarrhea, increased emotional lability, desire to eat salt, amenorrhea, loss of libido, impotence, cramps and muscle contractures.

Physical examination. Hypotension, hyperpigmentation of the skin, patches of dark gray pigmentation on the mucosa of the mouth, vagina and rectum; asymmetric areas of vitiligo, hair loss, dehydration.

Paraclinical examinations

The best direct test is stimulation with ACTH intravenously for 8 hours (administer oral dexamethasone to prevent reactions), determination of 17-hydroxysteroids and 17-ketosteroids and/or plasma levels of cortisol used as an index of adrenal response. The absence of response is obtained in both primary and secondary insufficiency.
To differentiate both forms, the administration of ACTH is continued for several days and the determination is repeated. If the levels increase, it orients to the secondary type. Determination of plasma cortisol and urinary 17-ketosteroid and 17-hydroxycorticosteroid levels has relative value.

Indirect indications of adrenal insufficiency are given by a group of cheaper and less complicated tests, although not as specific as the previous ones: fluid overload test, salt suppression test, measurement of the sodium-potassium ratio in blood and saliva, absolute eosinophil count, glucose tolerance test.

Other findings: normocytic normochromic anemia, lymphocytosis.
Hypoglycemia, hyponatremia, hypochloremia, hyperkalemia.

Other procedures:

Radiology of sella turcica, thorax (heart size), abdomen (calcifications).

Electrocardiogram. Signs of hypokalemia.

Electroencephalogram. Generalized theta activity, alpha rhythm suppression, occasionally delta activity.

For the diagnosis of the pituitary type, the association with other endocrine disorders is important; elevated cholesterol, low T3-T4, decreased urinary gonadotropins, oligospermia.

Endometrial biopsy.

Determination of ACTH and mepirapone test after prolonged administration of ACTH.

Syndromogenesis and etiology

Several mechanisms can induce chronic adrenal insufficiency. They are subdivided into two types: primary and secondary.

Primary type:

1. Primary atrophy (due to autoantibodies): in this type there is a frequent association with other endocrine insufficiencies (pancreatic, gonadal), but with adrenal insufficiency as the most prominent element.
2. Infections (tuberculous, viral, bacterial, fungal).
3. Infiltration due to leukemia, reticuloendotheliosis, lymphomas, metastases, amyloidosis.
4. Hemorrhage: trauma, infections, anticoagulant treatment, thrombocytopenia, thrombosis.
5. Congenital familial (rare form).
6. Induced by drugs that alter the normal metabolism of cortisol.

Secondary type:

1. Pituitary insufficiency due to tumor, necrosis, iatrogenesis (medications, surgery, radiotherapy), suppression of activity due to hypothalamic insufficiency to produce corticotropin-releasing factor.

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Addison, T.: Diseases of adrenal capsules. London Med. Gaz.O.S. 43(N.S. 8):517-518, 1849.

Farwleg, FE: Adrenal cortical insufficiency. In Eisenstein, A.B. (ed.). The Adrenal Cortex, Boston, Little Brown, 1967.

Turkington, R.W. and H.E. Lebovitz: "Extrarenal endocrine deficiencies in Addison's disease." Amer. J. Med., 43:499-507, 1967.

SUCROSE INTOLERANCE SYNDROME

Syndromography

Clínic

It affects both sexes. It appears when an exclusively milk diet is discontinued and carbohydrates are added to the diet. It is characterized by diarrhea after each meal.

A good clinical test is to suspend foods that contain sucrose, dextrins or starch, which eliminates postprandial diarrhea.

Syndromogenesis and etiology

Congenital; severe isomaltase deficiency, reduced maltase activity by the intestinal mucosa.

Literature

Milne, M.D.: "Hereditary abnormalities of intestinal absorption." Br. Med. Bull., 23:279-284, 1968.

Weijers. H.A.: J.H. Kamer: D.A.A. van der Mossel, et al.: "Diarrhea caused by deficiency of sugar-splitting enzymes." Lancet, 2:296-297, 1960.

WATER INTOXICATION SYNDROME

Synonymy

S. of excess water. S. of overhydration. Dilutional S. (see hyponatremic syndrome).

Syndromography

Clínic

In its acute form: decreased attention, strange behavior, confusion, aphasia, incoordination, apathy alternating with violent behavior, marked muscle weakness.

In the chronic form: slow accumulation of water, weakness, drowsiness, apathy, anorexia, nausea, vomiting, salivation, lacrimation, watery diarrhea, non-excessive sweating: progresses to behavioral changes, convulsions and coma.

Physical examination. Skin: hot, moist, easily godet edema. Muscle contractions, tendon hyporeflexia, late Babinski. Hemiplegia may develop. Signs of pulmonary edema, especially if there is previous heart disease.

Paraclinical examinations

Blood. Decreased hemoglobin and hematocrit, macrocytosis with little decrease in hemoglobin concentration. Low sodium, low potassium, normal or high in severe forms. Normal or low urea nitrogen except if it was previously increased. Low baking soda.

Urine. Variable volume, low specific gravity, low or normal sodium and chlorine.

Syndromogenesis and etiology

Administration of water that exceeds the renal excretion capacity. Potomania. Generally, excessive water administration is iatrogenic in patients with excessive ADH secretion, postoperative state with decreased renal flow, Addison syndrome, acute renal failure, congestive heart failure, abundant enemas in hyponatremic patients and children with megacolon.

Literature

Goldberger, E.: A primer of water, electrolyte, and acid-base syndromes, ed. 3, Philadelphia, Lea & Febiger, 1965.

KALLMAN SYNDROME

Synonymy

S. of olfactory-genital dystrophy. S. of olfactogenital dysplasia. S. of gonadotropic hypogonadism-abnormalities. S. of anosmia-eunochoidism.

Syndromography

Clínic

It affects both sexes with a predominance in the male in a 3:1 ratio, with the possibility of a female carrier. The body is proportionate.

It is characterized by hypogonadism: lack of secondary and primary sexual characteristics (penis, testicles and infantile-type scrotum); in the female: uterine and ovarian hypoplasia.

Eunochoidism. Anosmia (only in men).

Partial red and green blindness. Cleft lip and/or cleft palate. Shortening of the fourth metacarpal. Mental retardation at times.

Paraclinical examinations

Blood. Low gonadotropin values. Negative methopyrone test.

For some authors, gonadotropin levels are within the limits of normality, while others find them greatly decreased.

Urine. Marked decrease in 17-ketosteroids and gonadotropins.

Smell tests. Anosmia. They should be performed in all cases of hypogonadism.

Testicular biopsy. Marked inhibition of testis maturation with absence of Leydig cells.

Syndromogenesis and etiology

Hypoplasia of the olfactory lobe of the brain that induces hypothalamic-pituitary hypofunction, due to the lack of release factor (releasing factor) in hypothalamic gonadotropin.
This causes a lack of follicle and interstitial cell stimulating hormone and therefore spermatogenesis.

Anosmia is caused by aplasia of the olfactory bulb.

Female carriers have partial anosmia.

The gene called KALIG-1 has homology with molecules involved in cell adhesion and the axonal pathway, which supports the idea that this genetic mutation causes an underlying defect in neuronal migration.

The etiology is genetic and is transmitted in a recessive manner linked to the X chromosome with predominance in males.

Literature

Franco, B. et al.: "A gene deleted in Kallman's syndrome shares homology with neural cell adhesion and axonal path-finding molecules." Nature, 353:529-536, 1991.

Maester de San Juan, A.: Total lack of olfactory nerves, with anosmia in an individual in whom there was congenital atrophy.
p. 211, El Siglo Médico, Madrid, 1856.

Sparkes, R.S.; R.W. Simpson and C.A. Paulsen: "Familial hypogonadotropic hypogonadism with anosmia." Arch. Int. Med., 121:534-538, 1968.

LAUNOIS SYNDROME

Synonymy

Neurath-Cushing S. S. of gigantism with hypopituitarism. S. of partial hypopituitarism with acromegaloid infantile gigantism. S. of gigantism-adiposogenital dystrophy.

S. de Launois-Cléret.

Syndromography

Clínic

Adolescents with delayed skeletal growth, delayed puberty and muscle weakness. At approximately 18-20 years of age, abnormal growth occurs and continues until 27-30 years of age.

Physical examination. Pale, thin skin, youthful appearance, broad face, little body and facial hair, small penis and testicles, eunuchoid appearance, high-pitched voice; long limbs, feet and hands, no epiphyseal fusion. It could be accompanied by thyroid and adrenal insufficiency.

Paraclinical examinations

Blood. Determination of gonadotropins, 17-ketosteroids, l1-deoxycortisol metabolites, PBI and I-131 uptake

Radiology of the sella turcica. Normal or signs of tumor, bone age determination.

Computerized axial tomography of the sella turcica.

Syndromogenesis and etiology

Idiopathic or due to chromophobe adenoma, craniopharyngioma with decreased production of gonadotropic hormones and occasionally TSH and ACTH. Increase in the production of growth

Literature

Launois, P.B. and R. Roy: Etudes biologiques sur les Géants. Paris, Masson, 1904. p.50.

Launois, P.B. and M. Cléret: "Le syndrome hypophysiare adipose-genital. " Gaz. h'Hop., 83:57: 83. 1910.

Sarver, M.E.; G. Sabeh; G.H. Fetterman, et al: "Fractional hypopituitarism with gigantism and normal sella turcica." N. English J. Med., 271:1286-1 289, 1964.

LAUNOIS-BENSAUDE SYNDROME

Synonymy

S. of Madelung. S. de Buschke II. S. of symmetrical adenolipomatosis.

Syndromography

Clínica

It generally appears in men between 35 and 45 years of age. At first, the patient notices that he cannot button his shirt collar and discovers great diffuse swelling in the posterior region of the neck and the appearance of symmetrical submandibular masses and successively other lipomas in the chest and the rest of the body with the exception of the limbs.

Generally, there is asthenia and apathy. Compression of a peripheral nerve produces pain (sometimes paresis), dyspnea and cough may also appear.

Physical examination. Lipomas are round, soft, of variable size but always large or gigantic, not well defined, not adhered to superficial or deep planes. Cyanosis and exophthalmos may appear.

Paraclinical examinations

Biopsy.

Blood. Liver function tests were somewhat choppy, as was the lipidogram.

Endocrine study. There may be a slight increase in basal metabolism and occasional decrease in 17-ketosteroid excretion.

Syndromogenesis and etiology

Ectodermal anomaly.

The etiology is unknown.

Literature

Gate, A.; J.M. Robert: "Le syndrome de Launois-Bensaude (a propos de 12 cas cliniques)". Ann. Chir. Plast., 11:193-197, 1966.

Launois-Bensaude: Ládenolipomatose symetrique". Bull. Soc. Méd. Hop. Paris, 15:298-318, 1898.

Madelung,: Ueber den Fetthais (diffuses Lipom des Halses). Arch. Klin. Chir. Berlin, 37:106-130, 1888.

SYSTEMIC MULTICENTRIC LIPOBLASTOSIS SYNDROME

Synonymy

S. of multiple lipoma. S. of lipoblastic sarcoma with metastasis. S. of metastasizing lipoma.

Syndromography

Clinic

Variable according to the location of fatty tissue growth.

Paraclinical examinations

Biopsy of the lesion.

Biochemical study of lipids.

Syndromogenesis and etiology

Alteration of lipid metabolism of unknown cause nocida.

Literature

Tedeschi, C.G: "Systemic multicentric lipoblastosis." Arch. Path., 42:320-337, 1946.

Montgomery, H.: Dermopathology, New York, Hoeber, 1967.

PARTIAL LIPODYSTROPHY SYNDROME

Synonymy

Barraquer-Simons disease. S. of progressive lipodystrophy. S. of progressive cephalothoracic lipodystrophy. S. de Kobbekling-Dunnigan.

Syndromography

Clínic

It predominates in females 4:1. Symptoms appear between 5-15 years and are manifested by the symmetrical absence of facial fat with or without disappearance of fat in the arms, chest, abdomen and hips. Fat retention in lower limbs.

It has also been reported: hepatomegaly, mental retardation, albuminuria and pyelonephritis.

Syndromogenesis and etiology

Familial incidence is very rarely reported. The presence of abundant fat in some places and almost total loss in others is curious. It has been thought of as a diencephalic disorder.

The etiology is unknown. The review of the literature suggests the existence of two types: one confined to the limbs and another that affects the trunk.

An X-linked dominant inheritance with lethality in males and the existence of at least two allelic types has been suggested.

Literature

Kobberling, J. and M.G. Dunnigang: "Familial partial lipodystrophy: two types of an X linked dominant syndrome, lethal in the hemizygous state". J. Med. Genet., 23:120-127, 1986.

LEG LIPOEDEMA SYNDROME

Syndromnography

Clinic

Frequent family history of bulky legs. Exclusively in women. Emotional and physical disorders due to the appearance of the legs. Worsening of symptoms in warm weather.

Physical examination. Gradual, bilateral buttock and leg augmentation. Accumulation of fat and water sensitive to pressure, soft edema, but with little godet. Generally, fat accumulation is limited to the legs and the rest of the body is normal, although occasionally there is generalized obesity. The feet remain normal in size and configuration.

Syndromogenesis and etiology

Possibly it has a hereditary constitutional character.

There is an emotional reaction triggered by the fact that very thick legs are considered ugly in the modern world. Ancient civilizations and the Hottentots considered them a sign of beauty.

The etiology is unknown.

Literature

Allen, E.V. and E.A. Hines, Jr.: "Lipedema of the legs: A syndrome characterized by fat legs and orthostatic edema." Proc. Staff. Meet. Mato Clinic, 15:184-187, 1940.

Wold, L.E.; E.A. Hines and E.V. Allen: "Lipedema of legs: A syndrome characterized by fat legs and edema." Ann. Int. Med., 34:1243-1250, 1951.

LIPOGRANULOMATOSIS SYNDROME

Synonymy

S. of disseminated lipogranulomatosis. Farber's disease.

Syndromography

Clinic

It begins between 1 and 4 months after birth. Hoarse voice, anorexia, vomiting, low-grade fever, malnutrition, motor delay.

Physical examination. Generalized polyarthritis with antalgic flexion contractures with 1-2 cm subcutaneous granulomas in pressure and periarticular areas. Polyadenopathy; dyspnea due to lung injury, laryngitis.

Paraclinical examinations

Joint biopsy.

Culture of skin fibroblasts. With ceramidase activity of only 5% of normal and 50% in heterozygotes.

Syndromogenesis and etiology

Genetics, autosomal recessive. Accumulation of ceramides, due to acid ceramidase deficiency, in tendons, synovial membranes, larynx, viscera, central and autonomic nervous system.

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Literature

Dulaney, J.; A. Milunski: J.B. Sidbury, et al.: "Diagnosis of lipogranulomatosis (Farber's disease) by use of cultured fibroblasts." J. Pediat., 89:59, 1976.

Farber, S.: lipid metabolic disorder-disseminated ' lipogranulomatosis'. A syndrome with similarity to, and important difference from Nieman-Pick and Hand-Schuller-Christian disease", Am. J. Dis. Child., 84:499, 1952.

Rampini, S. und J. Clausen: Farbersche krankheit (disseminierte Lipogranulomatose). Klinisches Bild und Zusammentafassung der chemischer Befunde". Hlev. Paediat. Acta, 22:500, 1967.

NEUROVISCERAL LIPODOSE SYNDROME

Synonymy

S. of late infantile systemic lipoidosis. S. variant of the Hurler. Tay-Sachs diseases with visceral impact. S. by pseudo Hurler. S. of generalized gangliosidosis. Betagalactosidase deficiency syndrome.

Syndromography

Clinic

It affects both sexes. From an early age there is mental and motor retardation.

Physical examination. Abnormal facies, macroglossia (occasional), peripheral edema, ascites (occasional), hepatosplenomegaly, cherry red spots on the retina (occasional).

Paraclinical examinations

Skeletal radiology. Lesions similar to those observed in Hurler syndrome.

Blood and bone marrow. Vacuolated lymphomonocyte cells, Adler leukocyte anomalies, increased alkaline phosphatase.

Syndromogenesis and etiology

Excessive accumulation of G4 ganglioside and abnormal ceramidehexoside pattern. Difficulty in differentiating it radiologically from Hurler syndrome, hematologically from Niemann-Pick, and clinically from Tay-Sachs. Anatomical and laboratory findings make the differentiation.

The etiology is unknown.

Literature

Caffey, J.: "Gargoylism (Hunter-Hurler disease, dysostosis multiplex, lipochondrosdytrophy): prenatal and early postnatal bone lesions and their early postnatal evaluation." Bull. Hosp. Joint Dis., 12:38-66, 1951.

Landing. B.H.; IN. Silverman; J.M. Craig, et al.: "Familial neuro-visceral lipidosis." Amer. J. Dis. Child., 108:503-522, 1964.

Suzuki, K. and G.C. Chen: “Morphological, histochemical and biochemical studies on a case of systemic late infantile lipidosis (generalized gangliosidosis).” J. Neuropath. Exp. Neurol., 27:15-38, 1968.

LUBS SYNDROME

Synonymy

S. of familial male pseudohermaphroditism with labial testicles and partial feminization (see testicular feminization syndrome).

Syndromography

Clinic

Pseudohermaphrodite male. Signs vary with age.

Physical examination. Elongated clitoris, lips with scrotal characteristics containing the testicles, urogenital sinus containing the urethra. Female breast development and hair distribution.

Paraclinical examinations

Testicular biopsy.

Endocrine study. Absence of significant abnormalities. Sex chromatin: no chromatin-positive cells.

Chromosome study.

Syndromogenesis and etiology

Family occurrence. Possible lack of X chromosome dislocation.

Literature

Lubs, H.A.; 0. Vilar and D.M. Bergenstal: “Familial male pseudohermaphroditism with labial tests and partial feminization: endocrine studies and genetic aspects.” J. Clin. Endocr. Metab., 19:1110-1120, 1959.

Peris, L.A.: "Genetic causes of primary amenorrhea." Clin. Obstet. Gynecol., 11:835-853, 1968.

MALABSORPTION SYNDROME AND MACROAMYLASEMIA

Syndromography

Clínic

Apparently limited to mature-aged females. Chronic diarrhea, clinical symptoms of malabsorption syndromes.

Paraclinical examinations

Blood. Persistent amylasemia of the IS type. Low xylose absorption.

Stool. Very increased fat.

Syndromogenesis and etiology

The nature of the relationship between malabsorption and the presence of IS amylase is unknown. Patients with persistent macroamylasemia type 7S do not have malabsorption.

Literature

Levitt, M.D.; E.J. Goetzl MR. Cooperband: "Two forms of macroamylasaemia." Lancet, 1:957-958, 1968.

Wilding, P.: W.T. Cooke and G.I. Nicholson: “Globulinbound amylase: a cause of persistently elevated levels in serum.” Ann. Int. Med., 60:1053-1059, 1964.

MAURIA C SYNDROME

Synonymy

S. of juvenile diabetes with dwarfism, hepatomegaly and obesity.

Syndromography

Clínic

It develops slowly in fragile-looking diabetic children whose diabetes is difficult to manage, growth is slow.

Physical examination. Dwarfism, moon-faced obesity, hepatomegaly, hypersensitivity to rapid-acting insulin and good response to slow-acting insulin.

Paraclinical examinations

Evaluation of diabetes and pituitary and adrenal corticosteroid function. Rule out deposit diseases.

Syndromogenesis and etiology

Nutritional deficiency, insulin deficiency. Possible metabolic disorder due to diabetes or associated condition.

Bibliography

Guest, G.M.: "The Mauriac Syndrome." Diabetes, 2:415-417, 1953.

Mauriac, P.: "Gros ventre. Hépatommégalie. Troubles de la crissance cher les enfants diabétiques, traités depuis plusieurs annés par l'insuline." Gaz. Hebd. des Sci. Med. de Bordeaux, 51:402-404, 1930.

MAPLE SYRUP

Synonymy

S. de Menkes I. S. of branched-chain ketonuria.

Syndromography

Clínic

It affects both sexes, from the first week of birth and is manifested by vomiting, difficulty feeding, insufficient development, absence of the prehension reflex, and gasping breathing.

Later rigidity and opisthotonos. Hypoglycemic crises may occur. Severe mental retardation. Urine that smells like honey or maple syrup.

Paraclinical examinations

Blood. Investigation of the enzymatic activity of leukocytes showing a blockage of the metabolism of branched-chain keto acids. Increased plasma levels of leucine, isoleucine, valine, and presence of alloisoleucine. This amino acid is increased in urine and cerebrospinal fluid.

Syndromogenesis and etiology

It is due to the interruption of the metabolic pattern of branched-chain keto acids, due to a defect in the enzyme alpha-ketoacid-dehydrogenase (debranching), which is a multienzyme complex associated with the inner membrane of the mitochondria.

It is of genetic etiology and is transmitted by autosomal recessive inheritance.

Literature

Dancis, J. and M. Levitz: Maple syrup urine disease (branched chain ketonuria). In Stanbury, J.B., J.B. Wyngaarden and D.S. Frederickson: The metabolic basis of inherited disease. ed. 2. New York, McGraw-Hill, 1966.

Menkes, J.H.; P.L. Hurst and J.M. Craig: "A new syndrome: progressive familial cerebral dysfunction associated with an unusual urinary substance". Pediatrics, 14:462-466, 1954.

HPERPARATHYRODEAL MYOPATHY SYNDROME

Synonymy

Muscular S. in hyperparathyroidism.

Syndromography

Clínic

It is rare. Fatigue, muscle weakness appear (subjective weakness is a common symptom in primary hyperparathyroidism, but this syndrome is characterized by objective weakness and atrophy)

Physical examination. Objective muscle weakness, muscle atrophy that raises suspicion of primary myopathy.

Paraclinical examinations

Blood. Typical findings of hyperparathyroidism (see this one).

Urine. Increase in creatinine excretion.

Muscle biopsy.

Electromyogram. High percentage of potential polyphasic.

Syndromogenesis and etiology

It originates from a parathyroid adenoma.

Literature

Frame, B.; E.G. Heinze; M.A. Block, et al.: "Myopathy in primary hyperparathyroidism." Ann. Int. Med., 68:1022-1027, 1968.

Vicale, C.T.: The diagnostic features of a muscular syndrome resulting from hyperparathyroidism, osteomalacia, owing to renal tubular acidosis and perhaps to related disorders of calcium metabolism. Trans. Amer. Neurol. Ass., 74th Meeting, 1949, p. 143.

IODINE-INDUCED MYXEDEMA AND GOITRE SYNDROME

Syndromography

Clínic

It appears in individuals subjected to prolonged administration of iodides due to thyroid dysfunction or other causes. A diffuse goiter develops solitary or accompanied by myxedema and other symptoms of hypothyroidism.

Paraclinical examinations

Thyroid function tests. They show hypofunction. Rebound phenomenon when discontinuing iodides.

Syndromogenesis and etiology

Blockade of organic iodization due to prolonged administration of iodine or a basic sensitivity to iodine probably related to a pre-existing defect in thyroid metabolism.

Literature

Bell, G.O.: Prolonged ingestion of iodine in the pathogenesis of simple goiter and myxedema. Tr. Amer. Goiter A., 1952, p.28.
Oppenheimer, J.H. and H.T. McPherson: "The syndrome of iodide-induced goiter and myxedema." Amer. J. Med., 30:281- -288, 1961.

Thompson, W. O.; PK.Thompson; A.G. Brailey, et al.: "Myxedema during the administration of iodide in exophthalmic goiter." Amer. J. Med. Sci., 179:733-749, 1930.

MIXEDEMATOUS-CEREBELLAR SYNDROME

Syndromography

Clinic

Patients with hypothyroidism and signs of myxedema who present cerebellar symptoms (ataxia, incoordination), which appear simultaneously or years after the onset of myxedema.

Paraclinical examinations

See Basedow syndrome for evaluation of thyroid function.

Syndromogenesis and etiology

It is due to a deficiency of thyroid hormone. The pathogenesis of cerebellar symptoms is not well elucidated.

Literature

Jellinek, E.H. and R.E. Kelly: "Cerebellar syndrome in myxoedema." Lancet, 2:225-227, 1960.
White, E.W.: "Myxoedema associated with insanity." Lancet, 1:974-976, 1884.

MORGAGNI-STEWART-MOREL SYNDROME

Syndromography

Clinic

Almost exclusively in obese, hirsute women, with various neuropsychiatric symptoms. Internal frontal hyperostosis.

Paraclinical examinations

Skull radiology. Smooth, rounded endostoses, projecting into the cranial cavity, smaller than 1 cm and not extending beyond the coronal suture.

Syndromogenesis and etiology

Generalized metabolic disorder that produces abnormality of the inner table of the frontal bones of the skull.

NARCOLEPSY SYNDROME - DIABETOGENIC HYPERINSULINISM (FUNCTIONAL)

Synonymy

S. of metabolic obesity. S. of psychosomatic obesity. S. of euthyroid hypometabolism. S. of refractory hypothyroidism. S. of refractory chronic fatigue.

Syndromography

Clinic

Family history of narcolepsy (48%), irresistible drowsiness with inappropriate and pathological sleep (100%); cataplexy (50%) triggered by a sudden emotion, hypnotic hallucination (56%), paralysis during sleep (49%), inability to move when waking up or at the beginning of sleep. It is frequently associated with vascular headache (65%) (see Horton's headache), peripheral neuropathy especially of the lower limbs (34%), spontaneous leg cramps (37%) , (see Witmaak-Exbon syndrome), angina pectoris and arrhythmias (32%). Psychiatric disorders: anxiety and depression.

physical examination Obesity (75%), therapeutic response to analeptic agents (100%). Frequent association of: recurrent edema (50%), café au lait spots (24%), occipital nevus and gray hair.

Paraclinical examinations

Blood. Fasting blood glucose, glucose tolerance test, in the morning and afternoon; insulin determination; electrolytes, cholesterol, uric acid, protein-bound iodine (PBI).

Diuresis.

Respiratory functional tests.

Electrocardiogram.

Electroencephalogram.

Basal metabolism.

Pharmacological tests. Methylphenidate, insulin, tolbutamide and liothyronine.

Syndromogenesis and etiology

Hypoglycemia related to eating habits, narcoleptic hypokinesia, accelerated lipogenesis in relation to chronic hyperinsulinism, functional disorder of the nervous system.

bright

Literature

Roberts, H.J.: "Obesity due to the syndrome of narcolepsy and diabetogenic hyperinsulinism: clinical and therapeutic observations on 252 patients." J. Amer. Geriat. Soc., 15:721-743, 1967.

NELSON SYNDROME

Synonymy

S. of Nelaton. See addisonian syndromes.

Syndromography

Clinic

It occurs in patients with adrenal hyperplasia, 1-8 years (average 3 years) after adrenalectomy.

Physical examination. Hyperpigmentation of the skin and mucous membranes, decreased visual field, other neurological signs typical of a pituitary tumor.

Paraclinical examinations

Skull radiology. Signs of intrasellar tumor.

ACTH dosage.

Computerized axial tomography of the skull.

Syndromogenesis and etiology

After an adrenalectomy, a chromophobe tumor of the pituitary gland develops with excessive ACTH secretion.

Literature

Nélaton, A.: “Affection singuliere des os du pied.” Gaz. des Hop., 4:'3, 1852.

Nelson, D.H.; J.W. Meakin and G. W. Thorn: "ACTH-producing pituitary tumors following adrenalectomy for Cushing's syndrome." Ann. Int. Med., 52:560-569, 1960.

TYPEI MULTIPLE ENDOCRINE NEOPLASM SYNDROME

Synonymy

S. of MEN I. S. of Wermer.

Syndromography

Clínica

Rare in childhood or after age 60. It includes tumors or hyperplasia of: parathyroid, pancreatic islet cells, adrenal cortex and thyroid. Symptoms depend on the affected gland; About two-thirds of patients have adenomas of two or more systems and one-fifth of three or more systems. The majority of those affected present one of the following problems:

1. Peptic ulcer and its complications.
2. Hypoglycemia.
3. Hypercalcemia, nephrocalcinosis or both.
4. Pituitary symptoms: headache, visual field defects, secondary amenorrhea.
5. Multiple skin lipomas.
6. A minority, less than 10%: acromegaly, Cushing's, non-functional thyroid adenomas, hyperthyroidism, hepatomegaly due to metastasis, hot flashes due to carcinoid syndrome.

Paraclinical examinations

They must be selected according to the clinical manifestations. First-degree relatives should be studied and tested: calcium, phosphorus, prolactin and gastrin in serum, pancreatic polypeptides in serum.

Syndromogenesis and etiology

It has been considered that the basic anomaly resides in the islet cells, with their enormous capacity for synthesis of hormones, and changes in the other glands have been attributed to secondary effects of insular hormone hypersecretion.

Other theories classify this syndrome as a neurocrestopathy, which involves deficient differentiation or regulation of the embryonic neural crest, which is the primordium of at least part of the endocrine system. The endocrine components of the neural crest have been classified into a subsystem of APUD cells, named for their ability to uptake and decarboxylate amine precursors. There is not much data to support this hypothesis. It is more likely that cells of diverse origins develop similar characteristics, that is, they represent a structural-functional convergence.

MULTIPLE ENDOCRINE NEOPLASMY SYNDROME TYPE II

Synonymy

S. of MEN II or IIA. S. de Sipple.

Syndromography

Clinic

The age at diagnosis varies between 2 and 67 years and includes:

1. Pheochromocytoma (often bilateral and sometimes extra-adrenal).
2. Medullary thyroid carcinoma (palpable or silent).
3. Parotid hyperplasia (in about half of the cases).
4. Gliomas, glioblastomas and meningiomas may also occur.

Paraclinical examinations

Blood. Immunoassay of calcitonin in serum makes the diagnosis of medullary thyroid carcinoma but there is also ectopic production in breast, lung and islet cell tumors; In high-risk individuals, when baseline serum calcitonin concentrations are normal, measurement can be used after intravenous administration of calciumpentagastrin.

Syndromogenesis and etiology

It may be more directly related to abnormal development of the neural crest, since both the adrenal medulla and the parafollicular or C cells of the thyroid originate from this crest. However, it has not been proven that the parenchymal component of the parathyroids has this origin.

Medullary thyroid carcinoma is transmitted as an autosomal dominant character. Examination of cells from both medullary thyroid carcinoma and pheochromocytoma in MEN II using X-linked genetic markers has led to the conclusion that the hereditary defect produces several clones of abnormal cells; Tumors then develop from a second mutation in the abnormal clone, which explains the appearance of diverse clinical types.

TYPE II MULTIPLE ENDOCRINE NEOPLASM SYNDROME

Synonymy

S. of MEN III or IIB: S. of mucosal neuromas.

Syndromography

Clinic

More malignant evolution than MEN II and is characterized by the triad: medullary thyroid carcinoma, pheochromocytoma and notable dysmorphic signs.

Physical examination. Neuromas of the conjunctival, labial and buccal mucosa, and the mucous membranes of the tongue, larynx and digestive tract; enlarged corneal nerves, protruding lips, soft tissue prognathism and habit similar to Marfan syndrome with hypotonia, lax joints, kyphoscoliosis, genu valgum and pes cavus. It may present café au lait spots or a type of diffuse skin pigmentation along with cutaneous neurofibromas. There may be megacolon.

Syndromogenesis and etiology

MEN III and II are different entities. In III, parathyroid hyperplasia and the production of hormones other than calcitonin due to medullary thyroid carcinoma are rare. The average survival is 30 years and 60 in MEN I.

NIEMANN-PICK SYNDROME

Synonymy

S. of lipoidosis due to sterolsphingomyelin. S. of sphingomyelina reticuloendotheliosis. Essential lipoid histiocytosis S. S. of magnocellular metamorphosis of the lymph node.

Syndromography

Clinic

Three main clinical forms: classic, visceral and juvenile.

Classic (infantile) form: occurs in 85% of cases and is rapidly progressive.

Visceral form: takes different viscera but no abnormalities of the central nervous system are observed.

Juvenile form: moderate course, hepatosplenomegaly is not very large, and appears late, central nervous system manifestations appear after several years, cherry red spots absent or late appearing, long survival.

In the juvenile form, other varieties known as:

Nova Scotia Group (Crocker-Farber): prolonged course, neurological manifestations progress slowly until reaching severe weakness. In this group the appearance of jaundice is a notable fact.

Subacute form: appears one year after birth with slow development of the classic clinical picture. There is visceral infiltration of sphingomyelin but not in the brain. Death occurs between 4-10 years of age.

It affects both sexes equally. It can occur in any race, although 40% correspond to Jews. Generally, no alterations are detected at birth. Between 1-2 months, difficulties in development begin to be observed, as well as mental retardation that progresses to apathy and clumsiness.

Physical examination. Weakness that can reach marasmus. Globulous abdomen, hepatosplenomegaly. Hyperpigmented spots can be observed in areas of the skin, some bluish, xanthomas are rare. Fundus examination shows the typical cherry-red spot that makes prominence, located in the macula, as in Tay-Sachs syndrome.

Paraclinical examinations

Blood. Anemia, vacuolated leukocytes. Kampine-Brady Kanferf test on washed white blood cells: low level of enzymatic activity for the hydrolysis of sphingomyelin and glucocerebrosides. bone marrow: presence of typical cells.

Biochemical study of phospholipids.

Lymph node biopsy. Large, foamy, vacuolated cells (Pick cells) are seen (as in the liver, spleen, lung, and brain) and show positivity to the Smith-Dietrich reaction for phosphatides.

Syndromogenesis and etiology

The syndrome is caused by an alteration of sphingomyelin metabolism, fundamentally a deficiency or absence of sphingomyelinase, which produces an accumulation of sphingomyelin and cholesterol in the cells of different organs, especially the nervous system and the reticuloendothelial system.

It is transmitted by autosomal recessive inheritance with different phenotypic manifestations.

The etiology is genetic.

Literature

Niemann, A.: "Ein unbekanntes Krankheitbild." Jahrb. Kinderh. n.F., 29:1-10, 1914.

Pick, L.: "Uber die lipoidzellige Splenohepatomegalie Typus Nieman-Pick als Stoffwecheslerkrankung." Med. Klin. 23: 1483- -1488, 1927.

RESIDUAL OVARIAN SYNDROME

Syndromography

Clinic

In hysterectomized patients who have preserved one or both ovaries, a condition characterized by continuous or intermittent pelvic discomfort, occasionally dyspareunia, occurs.

Physical examination. A persistent pelvic mass is palpable and generally larger than 5 cm.

Paraclinical examinations

Ultrasound.

Syndromogenesis and etiology

Ovary that continues or attempts to continue functioning and presents: cyst, atresia, hemorrhagic follicle, corpus luteum, perioophoritis. Occasionally endometriosis and neoplasia.

Literature

Grogan, R.H.: “Reappraisal of residual ovaries.” Amer. J. Obstet. Gynec., 97:124-129, 1967.

FAMILIAR PERIODIC PARALYSIS SYNDROME

Synonymy

of Westphal. S. de Cavaré-Westphal. S. de Cavaré -Westphal- -Romberg. S. of hypokalemic paralysis.

Syndromography

Clinic

It begins between 7-20 years (90%), with predominance in males (12:1). It is characterized by prodromal symptoms: sweating, heaviness of the limbs, paresthesias. During the night or upon waking up, the crisis occurs: flaccid proximal paralysis of the trunk and limbs in an asymmetrical manner and without sensitivity disorders that generally respect breathing, swallowing and gesticulation (they can only move their head somewhat). This condition lasts 6-24 hours (occasionally a week) and repeats at greater or lesser intervals. Sometimes the crisis is accompanied by: migraine, eyelid ptosis, bradycardia, arrhythmia that may include premature ventricular beats, ventricular tachycardia and cardiac dilation. Patients with repeated and severe seizures develop persistent loss of strength and dystrophic changes in the muscle.

Treatment during crises consists of the administration of potassium chloride. Acetazolamide reduces its frequency. Vitamin B. and chlorothiazide have also been useful.

Paraclinical examinations

Blood. Hypokalemia, 2-3 mEq/L (in crisis).

Urine. Hypocaliuria (in the crisis), followed by potassium diuresis the next day.

Electrocardiogram. Signs of hypokalemia.

Electromyogram. Complete galvanic and faradic muscle inexcitability with lack of propagation of action potentials along muscle fibers.

Syndromogenesis and etiology

It is due to a disorder of carbohydrate metabolism during sleep, which causes paralysis due to potassium falling below 3 mEq/L due to a disorder of the membrane potential in the muscle. The triggering factors for the crisis are: stress, infections, carbohydrate-rich meals, rest after exhausting efforts and certain medications (adrenaline, ACTH, thyroid, licorice, insulin).

The etiology is genetic. It is transmitted in an autosomal dominant manner, with reduced penetrance and expressivity; sporadic in 25%.

Literature

Dubowitz, V.: Muscle disorders in children. Ed. Saunders, Philadelphia, 1878. p. 120.

Goldflam, S.: "Ueber eine eigentumliche form von periodische, familiaerer wahrschenlich autointoxicatorischer paralyse." Vienna. Med. Presse, 31:1418, 1890.

Musgrave, W.: A periodical country. The philosophical transactions and collections to the end of the year 1700. London, 2:1727.

HYPOCALIEMIC PERIODIC PARALYSIS SYNDROME

Synonymy

S. of familial hypokalemic periodic paralysis. S. of sporadic hypokalemic periodic paralysis.

Syndromography

Clinic

In the affected families, males predominate, as well as in sporadic cases. The age of onset ranges from

I year until the fifth decade (90% between 7 and 21 years old). Between attacks the patient is asymptomatic, occasionally there is a slight decrease in muscle strength. The syndrome can be preceded by migraine attacks for years.

Precipitating factors: stress (infection, surgery, emotions, cold, menstruation), abundant meals rich in carbohydrates, rest after intense exercise, drugs (epinephrine, ACTH, DOCA, thyroid, licorice). Prodromes: most seizures occur in the early hours of the morning upon rising; During the day, stiffness, cramps, paresthesias, irritability, intense thirst announce them, exercise can abort them. The crisis begins in the legs and extends to the entire body, flaccid paralysis; There may be dysphagia, difficulty in eye and respiratory movements, but the sensory system is little affected; occasionally vomiting and constipation.

There is recovery in hours, a day or a little later.

Physical examination. Between crises, the muscles appear to increase in size. During them they have less consistency and increase in circumference. Decrease or absence of deep tendon reflexes. In some cases there is bradycardia and cardiomegaly.

Paraclinical examinations

Blood. During the crisis there is leukocytosis with neutrophilia and eosinopenia followed by recovery by lymphocytosis and eosinophilia; Normal potassium outside of crises and low during them.

Urine. Potassium falls together with the fall of this in blood.

Syndromogenesis and etiology

It appears to be a potassium metabolism disorder, inherited on an autosomal basis, although there are sporadic cases and families with an X-linked inheritance pattern have been described, which suggests genetic heterogeneity.

Literature

Shaknovitch: On a case of intermittent paraplegia. Russk. Vratch ch., 32:537, 1882, abstracted in London M. Rec., 12:130, 1884, by V. Idelson, M.D.

Streeten, D.H.P.: Periodic paralysis. In: Stanbury, J.B., J.B. Wyngaarden, D.S. Frederickson: The metabolic basis of inherited disease, New York, McGraw-Hill, 1966, p.905.

PELLIZZI SYNDROME

Synonymy

S. pineal. S. of precocious puberty. S. of early [illegible] (See pineal tumor syndrome)

Syndromography

Clinic

It begins in childhood, accelerated increase in weight, height, muscles and development of sexual characteristics, frequently associated with a neurological syndrome due to pineal tumor.

Paraclinical examinations

Skull radiology. Occasionally calcification of the pineal.

Computerized axial tomography of the skull.

Determination of gonadotropins.

Syndromogenesis and etiology

It is usually associated with destructive tumors of the pineal gland. The mechanism of production of the symptoms is due to the suppression of a hypothetical pineal hormone that would inhibit gonadal development, or an indirect stimulation of the secretion of gonadotropins by pressure on the hypothalamus and pituitary gland.

The etiology is unknown.

Literature

Cohen, R.A.; R.J. Wurtman; J. Axelrod, et al,: "Some clinical, biochemical, and physiological actions of the pineal gland." Ann. Int. Med., 61:1144-1161, 1964.
Pellizi, G.B.: The epiphyseal syndrome "early macrogenitosomia". Riv. Italian. Neuropathy Catania, 3:193-207, 250, 1910-11.

PINEAL GONADAL SYNDROME

See pineal tumor syndromes.

Syndromography

Clinic

In young men (hypogonadism and pineal tumor have not been described in females). Delay or absence of the development of primary and secondary sexual characteristics with neurological syndrome caused by the pineal tumor.

Paraclinical examinations

Skull radiology.
Computerized axial tomography.
Gonadotropins.

Syndromogenesis and etiology

Increase in the secretion of a hypothetical pineal hormone that would inhibit gonadal development.
The etiology is unknown.

Literature

Cohen, R.A.; R.J. Wurtman; J. Axelrod, et al.: "Some chemical, biochemical, and physiological actions of the pineal gland." Ann. Int. Me., 61:1144-1161, 1964.
Kitay, J.I: "Pineal lesions and precocious puberty, a review." J. Clin. Endocrinol., 14:622-625, 1954.

PORPHYRIA SYNDROME

Syndromography

Because of the difficulties in naming the different syndromes that are grouped with this name, a classification based on clinical, biochemical and genetic aspects is proposed:

Erythropoietic form:

1. Günther syndrome. It is a congenital erythropoietic porphyria syndrome.
2. Erythropoietic porphyria syndrome.
3. Enteropathic coproporphyria.
4. Crosby syndrome.

Liver form:

1. Acute and chronic porphyria.
2. Swiss type. It is an intermittent porphyria syndrome.
3. Dean-Barnes syndrome. Varied porphyria syndrome.
4. Symptomatic porphyria syndrome.
5. Hepatic coproporphyria.
6. Symptomatic porphyrias.
7. Hepatoma porphyria.

Clinic

Porphyria is characterized by photosensitive skin with a tendency to hyperpigmentation. Hypertrichosis. Frequent intestinal cramps. Splenomegaly. Peripheral or central paresis or paralysis. Pink urine that fluoresces in ultraviolet light due to the presence of porphyrins.

Literature

Schmid, R.: The Porhyrias. In Stanbury, J.B., J.B. Wyngaarden and D.S. Fredrickson: The metabolic basis of inherited disease. ed. 2, New York, McGraw-Hill, 1966.

PORPHYRIA CUTANEA TARDA SYNDROME

Synonymy

S. of chronic hepatic porphyria.

Syndromography

Clinic

The term has been used by different authors with different meanings.

It was originally described by Waldenström to describe patients with late onset of porphyrinuria and photosensitivity without neurological or abdominal signs.

Waldenström recognizes different types of porphyria cutanea Tarda while other authors recognize it with the same meaning originally described.

In this syndrome, pinkish urine and hepatomegaly of a hard consistency appear.

Paraclinical examinations

Urine. Intense pink color (uroporphyrin and coproporphyrin I and II

Blood. High sideremia.

Syndromogenesis and etiology

Autosomal dominant inherited condition with incomplete penetrance. It is related to syphilis, hepatitis, alcoholism, cholecystopathy, drugs and contraceptives.

ERYTHROPOIETIC PORPHYRIA SYNDROME

Synonymy

S. of congenital porphyria. Gunther I. S. of congenital erythropoietic porphyria. S. of congenital photosensitive porphyria.

Syndromography

Clinic

It affects both sexes equally and has a wide racial distribution. It begins at birth or during the first year of life. Episode of reddish urine, a fact that persists with great daily and seasonal variations. If the child is exposed to

sun rays (mainly during the first year) a vesicular or blistering type rash appears in the exposed areas that heals slowly, leaving an eschar. There are no abdominal or neurological manifestations.

Physical examination. Acute bullous manifestations and scars of varying degrees from mild to very severe (mutilating). Hypertrichosis (thin and blond lanugo), mild chronic jaundice, erythrodontia (better visualization with ultraviolet light), splenomegaly (inconstant finding whose size may vary during the course of the disease), normal blood pressure.

Paraclinical examinations

Blood. Normocytic normochromic anemia, intermittent and almost never intense; elevated reticulocytes; normoblasts in periphery; decreased red blood cell survival. Bone marrow: erythropoietic hyperplasia. In studies carried out with fluorescent light microscopy in uncolored samples, normoblasts present intense fluorescence in nucleated red cells, which reaches its highest intensity in the nucleus and perinuclear area and lowest in some reticulocytes.

Medulogram. Erythroblastic hyperplasia with erythroblastophagy.

Urine. The color varies from pale pink to deep red, with a predominance of type I porphyrin and in small amounts of type III and coproporphyrin.

Stool. Large amounts of coproporphyrin type I, little of type III and increased fecal urobilinogen.

Syndromogenesis and etiology

Autosomal recessive hereditary condition, always in homozygotes with overproduction of porphyrin type I, limited to the erythrocyte system, due to a deficiency of the urogenous III cosynthetase that produces protoporphyrin III and not I.

The phenomenon is metabolic in nature and is due to a disorder in the biosynthesis of porphobilinogen; uroporphyrin is probably due to the lack of an isomertase and the disinhibition of a repressive system. This leads to the formation of pathological erythroblasts (porphyroblasts) and erythrocytes (porphyrocytes) that have a limited lifespan.

Pathological uroporphyrin produced in excess is responsible for photosensitization and progressive dermatosis.

The etiology is genetic.

Literature

Gunther., H.: Sitzungebericht miedershein, Gesellschsft, f. Natur- und Heilkunde, Bonn 1911

Gunther, H.: “Die Hamatoporphyrie”. Deutsch. Arch. Klin. Med., 105: 89-146, 1911.

Schmidt, R.: The Porphyrias. In Stanbury, J.B., J.B. Wyngaarden, and D.S. Frederickson,: The metabolic basis of inherited disease. ed. 2, New York, McGraw-Hill, 1966.

Schultz, JH.: Ein fall von Pemphigus leprosus complicirt durch Lepra visceralis. Thesis, Greifswald, 1874.

LATENT PORPHYRIA SYNDROME

Syndromography

Clínica

Latent porphyria occurs more frequently in men. There are vague dyspeptic symptoms and nervousness. Typical attacks of acute porphyria can occur spontaneously or after ingestion of medications such as: barbiturates, sulfonamides, estrogens.

Paraclinical examinations

Chromatographic analysis of urine. An increase in the excretion of d-aminolevulinic acid and/or porphobilinogen is demonstrated.

Syndromogenesis and etiology

It is a clinical form of Swiss type porphyria.

SYMPTOMATIC PORPHYRIA SYNDROME

Synonymy

S. of cutaneous porphyria. S. of tarda porphyria.

Syndromography

Clínica

The syndrome is insidious in nature, occurs in middle age and affects both sexes. There is a history of high alcohol consumption.

Vesicular lesions and ulcerations followed by depigmented scars in areas of exposed skin are described.

There is hyperpigmentation of skin exposed to the sun.

Hepatomegaly.

Paraclinical examinations

Urine. With high excretion of uroporphyrin and other ether-insoluble porphyrins.

Liver function tests. Abnormal.

Syndromogenesis and etiology

There is a disorder of porphyrin metabolism possibly secondary to cirrhosis of the liver.

The etiology is unknown.

Literature

Schmidt, R.: The porphyrias. In Stanbury, J.B., J.B. Wyngaarden, and D.S. Fredrickson: The metabolic basis of inherited disease. ed. 2, New York, McGraw-Hill, 1966.

Waldenström, J.: "The porphyrias as inborn errors of metabolism." Amer. J. Med., 22:758-773, 1957.

SWISS TYPE PORPHYRIA SYNDROME

Synonymy

S. de Waldenström. S. of acute intermittent porphyria. S. of hepatic porphyria.

Syndromography

Clinic

This syndrome affects both sexes. It settles in young adults. Presents abdominal symptoms: cramping, intermittent, recurrent, localized or generalized pain attacks. Constipation, occasionally severe vomiting. Crises last days or months. During them there may be a slight fever. There is weight loss, electrolyte imbalance with dehydration.

Neurological symptoms: polyneuritis, including cranial nerves, functional brain disorders, with motor and sensory disorders, psychotic manifestations.

Physical examination. The abdomen is soft, no peritoneal irritation, non-constant hypertension, tagchycardia during crises.

Paraclinical examinations

Blood. During the crisis there is leukocytosis, hyponatremia.

Urine. Excretion of porphobilinogen.

Syndromogenesis and etiology

It is due to an increase in the concentration of D-aminolevulinic acid (ALA) and porphobilinogen due to blocking their metabolisms.

It is a metabolic defect that is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Gunther, H.: “Die Haematoporphyrie.” Deutsch. Arch. Klin. Med., 105:89-146, 1911.

Schmidt, R.: The porphyrias. In Stanbury, J.b.; J.B. Wyngaarden and D.S. Frederickson,: The metabolic basis of inherited disease. ed. 2, New York, McHgraw-Hill, 1966.

ERYTHROPOIETIC PROTOPOPHYRIA SYNDROME

Synonymy

S. of Magnus.

Syndromography

Clinic

It affects both sexes. It appears between childhood and adolescence. The symptoms are variable.

Brief minutes after or later after sun exposure, intense pruritus, erythema and edema appear and disappear in 12-24 hours without leaving sequelae.

Except during the acute phase, symptoms are negative. Splenomegaly has been reported. There is erythrodontia.

Paraclinical examinations

Blood. Only increase in the concentration of protoporphyrins in circulating erythrocytes.

Stool. Increase in protoporphyrins.

Medulogram. Increase in fluorescence limited to the cytoplasm of erythrocytes.

Syndromogenesis and etiology

It is due to excessive production of protoporphyrins in the bone marrow and possibly in other organs.

It is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Magnus, I.A.; A. Jarrett; T.A.J. Prankerd, et al.: "Erythropoietic protporphyria: a new porphyria syndrome with solar urticaria due to protoporphyrinaemia." Lancet, 2:448-451, 1961.

Schmidt, R.: The Porhyrias. In J.B. Stanbury, J.B. Wyngaarden, and D.S. Frederickson,: The metabolic basis of inherited disease. ed. 2, New York, McGraw-Hill, 1966.

DEAN-BARNES SYNDROME

Synonymy

S. of variegated porphyria. S. of porphyria cutaneous, Tarda, hereditary. Mixed hepatic porphyria S. Genetic South African porphyria S.

Syndromography

Clínic

It affects both sexes. Manifestations in women are moderate and may be accentuated during pregnancy. The incidence in white South Africans is 3:1,000.

The clinical onset is difficult to specify due to the variability in the intensity and type of skin lesion.

Manifestations generally appear in the third decade of life:

1. Skin manifestations as the only finding in half of the patients, increased sensitivity to light mechanical trauma, erythema, edema, blisters that leave moderate scars when healed, hyperpigmentation with depigmented atrophic areas.

2. Abdominal and neurological manifestations identical to the Swedish type of porphyria (see this one). The attacks are generally precipitated by the ingestion of barbiturates and other drugs.

Paraclinical examinations

Urine and feces. Increase in the excretion of protoporphyrin and coproporphyrin in the feces of all patients (even asymptomatic or almost asymptomatic). During crises, large amounts of aminolevulinic acid, porphobilinogen and porphyrins are eliminated in the urine.

Blood. Electrolyte imbalance (excessive vomiting, fluid loss).

Syndromogenesis and etiology

Hereditary (heterozygous), metabolic disorder of porphyrin synthesis, defect most likely limited to the liver.

Literature

Barnes, HD.: "Further South African cases of porphyrinuria." South African J. Clin., Sc., 1:117-169, 1951.

Dean, G.: *Porhyria". Br. Med. J., 2:1291-1294, 1953.

ACUTE PSEUDOABDOMEN SYNDROME DUE TO CORTICOSTEROID SUPRESSION

Syndromography

Clinic

After stopping corticosteroids, high fever, sweating, and abdominal pain appear.

Physical examination. Absence of usual signs of adrenal corticoid insufficiency. There is no rigidity of the abdominal wall or significant changes in intestinal fluid sounds.

Diffuse or localized abdominal pain.

Syndromogenesis and etiology

It is a rare complication that is observed in patients who are discontinued from corticosteroid treatment.

prolonged. It is observed in patients with rheumatoid arthritis in whom the dose is reduced, in patients with ulcerative colitis after surgery, when steroids are suppressed. These patients respond dramatically to cortical steroids or ACTH but not to antibiotics.

The etiology is unknown.

Literature

Spelberg, M.A.: "Postcolectomy syndrome with fever and abdomen-
"minal pain simulating acute abdominal inflammation caused by steroid withdrawal." Amer. J. Gastroenterol., 46:214-22 1, 1966.
Sprechler, M.: "Erfaringer med glucocorticoidbehandlede patients en Kirurgisk afdeling Saertryk fra." Nord. Med., 64:1357, 1960.

PSEUDOCUSHING SYNDROME

Syndromography

Clinic

It is especially seen in young individuals with hyperphagia.

Physical examination. Truncular obesity, abdomen and thighs.
Purpuric stretch marks.

Paraclinical examinations

Urine. Increased cortisol (more than normal but less than in true Cushing's), increased excretion of hydrocortisone metabolites. Plasma cortisol at a lower normal level.

Tests with ACTH, methopyrone and adrenal suppression. normal,

Syndromogenesis and etiology

Possible psychological imbalance in patients with constitutional factors.

The etiology is unknown.

Literature

Paschkis, K.E.; A.E. Rakoff: A. Cantarow, et al.: clinical Endocrinology. ed. 3, New York, Harper Row, 1967.

PSEUDOEUNUCODYSM SYNDROME

Synonymy

S. of fertile eunuchoidism.

Syndromography

Clínic

Absence of libido or sexual potency. Erections are
rare

Physical examination. Obesity, dry skin, poor axillary and pubic hair development. Absence of beard. Short penis, well-formed scrotums, normal-sized testicles, small prostate.

Paraclinical examinations

Sperm examination. Decrease in the number of sperm.

FSH. Normal.
LH. Decreased or normal. 17-ketosteroids markedly decreased.

Testicular biopsy. Few or absent Leydig cells. In the tubules, normal or reduced number of sperm.

Syndromogenesis and etiology

A primary or secondary androgenone deficiency arises.

Bibliography

McCullagh, E.P.; J.C. Beck and C.A. Schaffenburg: “A syndrome of eunuchism with spermatogenesis, normal urinary FSH and low or normal ICSH (fertile eunuchs).” J. Clin. Endocr., 13:489-509, 1953.

Nathanson, I.; L.E Towne and J.C. Aub: Normal excretion of sex hormones in childhood." Endocrinology, 28:851-865, 1941.

PSEUDOHYPERPARATHYRODISM SYNDROME

Syndromography

Clinic

Short stature, short metacarpals and metatarsals, round facies, ectopic calcification. Hypothyroidism or hyperthyroidism, saccharine diabetes, ovarian failure.

Paraclinical examinations

Blood. Elevated paratormone concentrations with normocalcemia or hypocalcemia; Variable deficiency of all pituitary hormones, including prolactin.

Syndromogenesis and etiology

Condition with frequent polyglandular manifestations. Heterogeneous syndrome, perhaps dominant inheritance linked to the X chromosome.

PSEUDOHYPOPARATHYROIDISM--ALBRIGHT IV SYNDROME

Synonymy

S. de Martin-Albright. St. of Albright IV. (See also pseudohypoparathyroidism

Syndromography

Clinic

Headache, weakness, lethargy, obtundation, paresthesia, dyspnea, laryngeal stridor, photophobia and blurred vision. Muscle cramps, abdominal pain, seizures. Some degree of mental deficiency.

Physical examination. Short stature, round face, thick neck, short limbs in relation to the trunk. Hands: thick, short and thick fingers, short metacarpal and metatarsal bones. Positive Chvostek and Trousseau signs.

Paraclinical Examinations

Blood. Normal serum phosphatase activity, low calcium, high phosphate.

Urine. Decreased excretion of calcium and phosphate.

Ellsworth-Howard test. There is no response of the kidney to parathyroid hormone.

Radiology. Early epiphyseal closure, subcutaneous and basal ganglia calcifications of the brain.

Syndromogenesis and etiology

Hereditary condition with decreased end-organ response (Seabright-Bantam syndrome) to normally secreted parathyroid hormone.

Literature

Albright, F.; C.H. Burnett; PH. Smith, et al.: *Pseudo-hypopa rathroidism-example of "Seabright-bantam syndrome"; report of three cases". Endocrinology, 30:922-932, 1942.
Paschkis, K.E.; A.E. Rakoff; A. Cantarow, et al.: clinical endocrinology. ed. 3, New York, Harper & Row, 1967.

PSEUDOHYPOPARATHYRODISM--ALBRIGHT II SYNDROME

Synonymy

S. of Albright II. S. of cerebro-metacarpo-metatarsal dystrophy.

Syndromography

Clinic

All signs of hypoparathyroidism: short stature, round face, sunken bridge of the nose, obesity, mental retardation, short fingers, positive " knuckle sign." Cataracts, abnormal dentition.

The name Seabright-Bantam has been used as an eponym for pseudohypoparathyroidism syndrome because in the Seabright-Bantam rooster, the tail feathers resemble the plumage of the female, and it represents an organic failure to respond to a specific stimulation, in this case: the androgen hormone.

In pseudohypoparathyroidism syndrome there is an insufficient response of biochemical changes with the administration of parathyroid hormone. This deficit in the response of a target organ to hormonal stimulation is also observed in other syndromes:

1. ADH-resistant diabetes insipidus syndrome (see this one).
2. Morsier syndrome.
3. Dwarfism (Seabright-Bantam type).
4. Savage syndrome (see this one).
5. Testicular feminization syndrome (see this one)

Paraclinical examinations

Blood and urine. Normality of calcium values and phosphate.

Radiology. Intracranial calcifications are observed, and in soft tissues, with ossification. Metacarpus, metatarsus and phalanges deformed or disproportionately short. Premature fusion of the conical epiphyses. Other alterations: arched long bones, osteoporosis, osteosclerosis, coxa vara or valga, syndactyly, osteochondromas.

Syndromogenesis and etiology

Probable anomaly linked to chromosome

The etiology is unknown.

Literature

Albright, F.; C.H. Burnett; P.H. Smith, et al.: "Pseudo-hypoparathyroidism-example of Seabright-Bantam syndrome"; report of 3 cases". Endocrinology, 30:922-932, 1942.
Albright, E; A.P. Forbes and P.H. Henneman: "Pseudo-pseudohy poparathyroidism." Trans. Ass. Amer. Physic., 65:337-350, 1952.
Barter, FC.: Parathyroid. In Beeson, P.B., and W. McDermott, eds.: Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia, Saunders, 1967.

REIFENSTEIN SYNDROME

Synonymy

S. of hereditary familial hypogonadism.

Syndromography

Clinic

It manifests itself with a clinical picture of male pseudohermaphroditism with hypospadias of variable severity and little evidence of secondary sexual characteristics. Postpubertal testicular atrophy with azoospermia. Gynecomastia occurs during puberty and persists with age.

It behaves with a phenotype: absent Müllerian ducts, Wolffian ducts with variable development, absent spermatogenesis, variable urogenital sinus from male to female, external genitalia with incomplete male development. female breasts.

Paraclinical examinations

Blood. Increased androstenedione and estrogen, as well as follicle-stimulating hormone. The concentration of testosterone in the blood is elevated.

Karyotype. 46,XY.

Syndromogenesis and etiology

The underlying defect is found at the level of androgen action and not its synthesis. Cytoplasmic deficiency of the dihydrotestosterone receptor has been detected.

Literature

Greiffin, J.E. and J.D. Wilson: "The syndromes of androgen resistance." New Eng. J. Med., 302:198-209, 1980.
Reifenstein, E.C., Jr.: "Hereditary familial hypogonadism." J. Clin. Endocr., 5:367, 1945.
Saez, J.M.; E. Perretti, et al.: "Familial male pseudoher maphroditism with gynecomastia due to a testicular 17-ketosteroid reductase defect." J. Clin. Endocr., 32:604, 1971.

RÉNON-DELILLE SYNDROME

Synonymy

S. of acromegaly with thyroid and ovarian insufficiency. (See Launois syndrome.)

Syndromography

Clinic

Hypotension, tachycardia, hyperhidrosis, heat intolerance, insomnia, oliguria.

physical examination The characteristics of acromegalic syndrome.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Rénon, L. and A. Delille: “Insuffisance thyro-ovarienne et hyperactivité hypophysaire (troubles acromégaliques).
Amélioration de lacromegalie par la medication hypophysaire" Bull. et Mem. Soc. Méd. Hop. of Paris, 3. s. 25:973-979, 1908.

GROWTH RETARDATION SYNDROME--RENAL AMINOACIDURIA-COR PULMONALE

Syndromography

Clínic

The growth rate begins to be slow from the first year of age, despite an apparent state of normal assimilation. The child presents slow motor development, difficulty walking due to muscle hypoplasia.

Frequent respiratory infections with cough and dyspnea, and development of cor pulmonale. Mental status is normal.

Physical examination. Delayed growth, reduced muscle and adipose mass, physical respiratory signs due to repeated infections, with pneumonia, bronchopneumonia, right ventricular hypertrophy.

Paraclinical examinations

Blood. Increased free fatty acids, normal amino acid level, elevated pCO, decreased oxygen saturation.

Polyglobulia.

Urine. High aminoaciduria, with high excretion of alpha-aminonitrogen (serine, threonine, histidine, lysine).

Electrocardiogram. Right ventricular hypertrophy.

Radiology. Osteoporosis. Normal kidneys.

Syndromogenesis and etiology

Genetic condition without established pattern, possible partial autosomal recessive hereditary transmission. Other family members may present with aminoaciduria without other clinical findings.

Defective function of the proximal renal tubules.

The etiology is unknown.

Literature

Rosenberg, L.E.; P.S. Mueller and D.M. Watkins: “A new syndrome:
familial growth retardation renal aminoaciduria and cor pulmonale II". Amer. J. Med., 31:204-215, 1961

Rowley, P.T.: P.S. Mueller: D.M. Watkin, et al.: "Familial growth retardation, renal aminoaciduria and cor pulnmonale." Amer. J. Med. 31:187-204, 1961.

SACCAROPINURIA SYNDROME

Syndromography

Clínic

Mental retardation, short stature, no seizures.

Paraclinical examinations

Urine. Excretion of large amounts of lysine, citrulline, histidine and saccharopine.

Blood. Sacaropine and citrulline, present in small quantities; lysine 4-5 times above normal and similar increase for citrulline.

Cerebrospinal fluid. High concentration of saccharopine.

Syndromogenesis and etiology

This syndrome is caused by an abnormal lysine metabolism of unknown cause.

Literature

Carson, N.A.J.; BB Scally; D.W. Neill, et al.: "sSaccharopinuria: a new inborn error of lysine metabolism." Nature, 218:679, 1968.

SCHEIE SYNDROME

Synonymy

S. of mucopolysaccharidosis V. S. variant of the Hurler.

Syndromography

Clínica

It affects both sexes. Visual impairment, normal intelligence, cases with psychosis have been reported.

Physical examination. Joint stiffness, claw hand, carpal tunnel syndrome, wide mouth, opacification of the cornea, uniform in early stages and later, harder towards the periphery. Retinitis pigmentosa. Normal or below normal height, excessive body hair. Aortic insufficiency.

Paraclinical examinations

Urine. Excess dermatan and heparan sulfate.

Radiology. Occasionally ribs in the form of "spaulae" and vertebral deformations. Carpal tunnel narrowing.

Syndromogenesis and etiology

Mucopolysaccharide metabolism disorder.

It is transmitted by autosomal recessive inheritance. He is allelic with Hurler syndrome. McKusick proposes that Hurler and Scheie be classified as mucopolysaccharidoses: I-Hel Hurler syndrome: I-S Scheie syndrome.

The etiology is genetic and is transmitted by autosomal recessive inheritance.

Literature

Scheie, H.G., GW., Hambrick, Jr. and L.A. Barness: “A newly recognized form fruste of Hurler's disease (gargolism)". Amer. J. Ophtahl., 53:753-769, 1962.

Williams, H.E.: "Heritable disorders of mucopolysaccharide metabolism." Qual. Med. 106:306-311, 1967.

SCHMIDT SYNDROME (M.B.)

Synonymy

S. of thyroid-adrenocortical insufficiency.

Syndromography

Clínica

It begins at any age, predominantly in women. Manifestations of hypothyroidism may precede or

follow those of Addison syndrome. Diabetes can be associated and its presence can precede or follow the manifestations of thyroid and adrenal insufficiency.

Paraclinical examinations

Study of antithyroid and antisupra antibodies

kidneys.

The other tests are the same as for hypothyroidism and hypoadrenalism.

Syndromogenesis and etiology

It is considered an autoimmune mechanism and commonly antibodies against thyroid, adrenals and possibly pancreas.

The etiology is unknown.

Literature

Carpenter, C.C.; N. Solomon; S.G. Silverberg, et al.: "Schmidt's syndrome (thyroid and adrenal insufficiency): A review of the literature and a report of fifteen new cases including ten instances of co-existing diabetes mellitus." Medicine, 43:153-180, 1964.

Schmidt, M.B.: "Eine biglandulare Erkrankung (Nebennieren und Schilddrusse) bei Morbus Addisonii." Verh. Deutsch. path. Ges. 21:212-22 1, 1926.

SCHROEDER I SYNDROME

Synonymy

S. endocrine-hypertensive.

Syndromography

Clínic

It predominates in the female sex, it is rare to see it in the male. It is characterized by obesity that appears relatively suddenly during menarche, menopause, or after multiple pregnancies or gynecological operations. Headache, irregular menstruation, rejection of salty foods. Tendency to easily appear bruises and ecchymosis.

Physical examination. Central type obesity with pale stretch marks on the thighs, noderate hirsutism. Arterial hypertension that appears after weight gain, changes in the fundus of the eye. The symptoms can be confused with Cushing's syndrome, maternal obesity syndrome or diabetic syndrome with macrofetus.

Paraclinical examinations

Blood. Moderate polycythemia, abnormal carbohydrate metabolism, abnormally low sodium and chloride concentrations.

Syndromogenesis and etiology

Dark. There may be a predisposing hereditary factor.

Literature

Schroeder, H.A.; D.F. Davies and H.E. Clark: "A syndrome of hypertension, obesity. Menstrual irregularities and evidence of adrenal cortical hyperfunction." J. Lab. Clin. Med., 34:1746, 1949.

Schroeder, H.A. and D.F. Davies: Studies on essential hypertension: V. An endocrine hypertensive syndrome". Ann. Int. Med., 40:516-539, 1954.

SYNDROME OF INADEQUATE ANTIDIURETIC HORMONE SECRETION

Synonymy

Schwartz-Bartter S. S. of cerebral hyponatremia with inadequate secretion of antidiuretic hormone. S. of cerebral salt loss. S. of inappropriate secretion of ADH.

Syndromography

Clinic

Headache, confusion, disorientation, hostility and other mental disorders with motor or sensory defect.

Physical examination. Normal reflexes, no dehydration, no edema, no signs of liver, kidney or heart disease.

Paraclinical examinations

Blood. Hyponatremia, moderate or severe. Normal urea nitrogen.

Urine. Hypertonic, with increased excretion of chlorine and sodium. Little response of hyponatremia to parenteral sodium solutions.

Radiology. Signs of trauma, tumor, infection.

Lumbar puncture.

Electroencephalogram.

Syndromogenesis and etiology

This syndrome is associated with lung carcinoma or is a consequence of brain injuries: trauma, tumors, vascular disease, encephalitis, meningitis, tuberculosis. It is the result of an electrolyte imbalance caused by hyponatremia resulting from inadequate secretion of ADH due to the aforementioned brain causes or due to the production of a substance with an effect similar to ADH.

Literature

Leaf, A.:; FC. Bartter: R.E. Santos, et al.: "Evidence in man that urinary electrolyte loss induced by pitressin is function of water retention." J. Clin. Invest.. 32:868-878. 1953.

Schwartz, W.B.; W. Bennett; S. Curelop, et al.: "A syndrome of renal sodium loss and hyponatremia probably resulting from inappropriate secretion of antidiuretic hormone." Amer. J. Med., 23:259-542, 1957.

SHEEHAN SYNDROME

Synonymy

S. of postpartum pituitary necrosis. S. of postpartum pituitary insufficiency. S. de Reye-Sheehan. S. of Simmonds- - Sheehan. (See Simmonds syndrome.)

Syndromography

Clínic

It appears postpartum after an acute initial shock. Failure to lactate, asthenia, hypoglycemic crisis, amenorrhea or menstrual irregularity. Anorexia.

Physical examination. Paleness disproportionate to anemia, loss of axillary and pubic hair, dryness of the skin, atrophy of the vaginal mucosa, slowing of the return phase of the tendon reflex. Bradycardia, hypotension, weight loss.

Paraclinical examinations

Basal metabolism. Low.

Blood. High cholesterol. Spontaneous hypoglycemia.

Iodine bound to protein. Low.

T3-T4. Bass.

Urine. Low excretion of corticosteroids, estrogens and pituitary gonadotropins.

Syndromogenesis and etiology

The syndrome is caused by thrombosis of the infundibular arteries of the portal pituitary system, by disseminated intravascular coagulation with shock during a complicated delivery. These alterations cause subsequent necrosis of the adenohypophysis, which results in secondary thyroid, adrenal corticosteroid and ovarian insufficiency. Precisely during pregnancy there is a pituitary hyperfunction that causes any ischemic process at that time to have a greater impact on the portal venous system of the anterior pituitary.

Literature

Sheehan, H.L. "Post-partum necrosis of the anterior pituitary". J. Path. Bact., 45:189-214. 1937.

Simmonds, M.: "Ueber Hypophysis-schwund mit todlichem Ausgang. Deutsch. Med. Wschr., 40:322-323, 1914.

STERILE MALE SYNDROME

Syndromography

Clinic

Phenotype with absent Müllerian ducts, male Wolffian ducts, absent or decreased spermatogenesis, male urogenital sinus, male external genitalia, generally male breast.

Paraclinical examinations

Blood. Normal or elevated testosterone, estrogen and LH production.

Syndromogenesis and etiology

Male pseudohermaphroditism due to a defect in the action of androgens. Perhaps recessive inheritance linked to chromosome X.

WILLIAMS SYNDROME

Synonymy

S. of idiopathic hypercalcemia of childhood.

Syndromography

Clínic

Supravalvular aortic stenosis, mental retardation, facies elf.

Paraclinical examinations

Blood. Hypercalcemia.

Syndromogenesis and etiology

Rare developmental disorder, of congenital origin, due to a mechanism of abnormal sensitivity to vitamin D and an increase in the circulating concentration of 1,25(OH)2 D, which reaches levels between 150 and 500 pg/mL. Children become hypercalcemic due to excessive intestinal absorption of calcium. The syndrome was first observed in England, after fortifying milk with vitamin D. It is not seen after the first year of life.

RELATED ENDOCRINOMETABOLIC SYNDROMES

Amenorrhea-galactorrhea syndromes

Inappropriate lactation. It includes four variants:

1. Ahumada del Castillo.
2. Chiari-Frommel.
3. Forbes-Albright.
4. Amenorrhea-galactorrhea-hypothyroidism.

Syndromes with amenorrhea-hirsutismno-obesity

Adrenogenital.

Cushing.

Constitutional hirsutism.

Stein-Leventhal.

Virilizing ovarian tumor.

Amino acid metabolic syndromes (inherited)

Isovaleric acidemia.

Abderhalden-Kaufman-Lignac.

Albinism.

Allan-Dent.

Carnosinemia.

Cystathioninuria.

Cystinuria.

Citrullinemia.

Cochrane.

Fanconi-De Toni.

Fanconi (adult type).

Folling.

Galactosemia.

Glycoglycinuria.

Hartnup.

Hyperamoniemia.

Hyperglycinuria-hyperglycinemia.

Hyperlysinemia.

Hypervalinemia.

Hypophosphatasia.

Histidinemia.

Lysine intolerance.

Joseph.

Leucinosis.

Lightwood.

Lignac-Fanconi.

Lowe.

Luder-Sheldon.

Methylmalonic.

Paine.

Growth retardation-renal aminoaciduria-corpulmonale.

Sacharopinuria.

Maple syrup (mapple syrup).

Hereditary tyrosinemia-tyrosinuria.

Neonatal tyrosinemia.

Tyrosinemia-tyrosinuria.

Wilson.

Syndromes with pituitary dwarfism

Anosmia-eunuchoidism.

Isolated growth hormone deficiency.

Multiple hormonal deficiency of the pituitary gland.

Dwarfism (Seabright-Bantam type).

Larin-Levi.

Enzyme deficiency syndromes

Hereditary alactasia.

Dormandy.

Sucrose intolerance.

Hereditary lactose intolerance.

Glucose-galactose malabsorption.

Syndromes with gonadal dysgenesis

XO Turner gonadal dysgenesis (XO type)

Swyer (type X0).

Rössle (type X0).

Gordan-Overstreet (type X0?).

II. XX gonadal dysgenesis

Turner (type XX/XO).

Swyer (type XX and XX/X0).

Rössle (type XX familial ovarian dysgenesis).

Stein-Leventhal (type XX and XXX).

Ovarian hypogenesis.

Prepubertal menopause.

II. XY Swyer gonadal dysgenesis (XY type)

XY gonadal agenesis.

Turner (male XY/XO type).

Klinefelter-Reinfenstein-Albright (type XXY or XXXY).

Stein-Leventhal (type XXY).

Hairless woman (XY type).

Others

Heller-Nelson.

Pseudo Klinefelter.

Noonan.

Kallmann.

Olfaction-genital.

Del Castillo.

Leydig hypogonadism.

Syndromes that break carbohydrate bonds

Hereditary alactasia.

Hereditary lactose intolerance.

Sucrose intolerance.

Dormandy.

Glucose-galactose malabsorption.

Glycogen storage syndromes

Andersen.

Forhes.

Hers.

Mc Ardle-Schmid-Pearson.

Pompe:

Cardiomegalic variant.

Acidomaltose deficiency variant.

Generalized variant.

Muscular variant.

Von Gierke.

Syndromes with hyposmolality

Addisonian.

Acute diabetes.

Hyponatremia.

Hyponatremia in the postoperative period.

Chronic cellular hyposmolality.

Inappropriate antidiuretic hormone secretion.

Syndromes with hypoparathyroidism

PRIMARY HYPOPARATHYROIDISM

Familiar:

Addison's-associated juvenile hypoparathyroidism (autosomal).

Hypothyroidism-juvenile-isolated-sex-linked (recessive).

Say George.

Hypoparathyroidism:

Acquired adult.

Acquired youth.

Juvenile-isolated-autosomal recessive.

Hydroelectrolyte-acid-base syndromes

WATER LOSS

Water depletion.

Diabetes insipidus.

ADH-resistant diabetes insipidus (see)

Hypertonicity due to solute overload.

EXCESS WATER

Water intoxication.

SODIUM LOSS

Addisonian.

Acute diabetes.

Hyponatremia.

Chronic dilutional hyponatremia.

Hyponatremia in the postoperative period.

Inappropriate antidiuretic hormone.

Water intoxication.

Mucoviscidosis.

Schroeder I.

Thorn.

METABOLIC ACIDOSIS Deficient bicarbonate.

Bywater.

Lightwood.

Acute diabetes.

Poisonous salicylate.

Hyperkalemic.

RESPIRATORY ALKALOSIS

See hyperventilation syndromes.

METABOLIC ALKALOSIS

Conn.

Hypokalemia.

Hypercalcemic.

Hypomagnesemia.

Hypermagnesemia.

Syndromes with lipid metabolic disorders

Bassen-Kornzweig.

Batten-Mayon.

Burger-Grutz.

Fabry.

Gaucher.

Hand-Schüller-Christian.

Familial hyperbetalipoproteinemia.

Familial hyperbeta-prebetalipoproteinemia.

Mixed hyperlipidemia.

Familial hyperprebetalipoproteinemia.

Metachromatic leukodystrophy.

Nieman-Pick.

Tangier.

Tay-Sachs.

Thiébaut.

Van Bogaert-Scherer-Epstein.

Wolman.

Lipodystrophic syndromes

Laurence-Siep.

Leiguel-Lavastine-Viard.

Gower's Panatrophic.

Weir-Michel.

Syndromes with mucopolysaccharidosis

Type I. Hurler.

Type II. Hunter.

Type II. Sanfilippo.

Type IV. Morquio.

Type V Scheie.

Type VI. Maroteaux-Lamy.

Type VII. Sly, Quinton, McAlister (beta-glucuronidase deficiency)

Chondroitin-4-sulfate-mucopolysaccharidosis (Thompson, Nelson, Castor).

Athetosis-keratasulfaturia (Maroteaux).

Transitional forms and mixed forms with mucolipidosis (Austin, Landing, mannosidosis, mucolipidosis I, Leroy).

Polyendocrine syndromes

Carcinoid in non-carcinoid malignant tumors.

Cushing's disease and hyperthyroidism.

Familial hyperparathyroidism.

Hyperparathyroidism-hyperthyroidism and thyroid cancer.

Multiple ectopic hormone production in a single neoplasm non-endocrine

Schenudt.

Sipple.

Wermer.

Zollinger-Elison.

Syndromes with progeria

Hutchinson-Gilford.

Gottron I.

Werner.

Metagery.

Seip-Lawrence.

Souques-Charcot.

Bamatter-Franceschetti-Klein.

Syndromes with pseudohermaphroditism

I. Female pseudohermaphroditism.

Congenital adrenocortical hyperplasia (congenital adrenogenital)

Virilizing adrenal corticoadrenal tumors (acquired adrenogenital).

Virilization during pregnancy due to exogenous causes (hormonal treatment) or endogenous causes (hormonal tumor of the mother).

II. Male pseudohermaphroditism.

Total feminization (testicular feminization: hairless woman).

Female type hair with pseudovaginal scrotal hypospadias.

Congenital adrenocortical hyperplasia.

Male tubular pseudohermaphroditism.

Boczkowski.

Reifenstein.

Swyer.

Gordon-Overstreet.

Irido-dentary (Weyers).

Syndromes with male pseudohermaphroditism

I. Defects in testosterone synthesis.

20-22 desmolase deficiency.

3-beta-hydroxysteroid dehydrogenase deficiency.

17-alpha-hydroxylase deficiency.

17-20 desmolase deficiency.

17-beta-hydroxysteroid dehydrogenase deficiency.

II. Defects in the action of androgens.

5-alpha-reductase deficiency.

Receptor disorders:

Complete testicular feminization syndrome.

Incomplete testicular feminization syndrome.

Reifenstein syndrome.

Infertile male syndrome.

Resistance with positive receptors.

III. Defects in Müllerian regression.

Persistent Müllerian duct syndrome.

IV, True hermaphroditism.

Lateral or alternating hermaphroditism (testicle on one side, ovary on the other).

Unilateral hermaphroditism (testis or ovary on one side and ovotestis on the other).

Bilateral hermaphroditism (ovotestis on both sides).

Mixed gonadal dysgenesis (testis on one side, udimentary ovary on the other).

Syndromes with tyrosinemia-tyrosyluria

Hepatic tyrosyluria (secondary to severe acute or chronic injury) unique in liver diseases).

Hereditary tyrosinemia-tyrosyluria.

Acute liver failure.

Baber:

with acute renal tubular dysfunction.

with S. de Fanconi.

Neonatal tyrosinemia.

Primary hypoparathyroid syndromes

Isolated infantile hypoparathyroid, linked recessive familial to sex.

Familial autosomal juvenile hypoparathyroidism associated with Addison.

Di-George.

Isolated autosomal recessive juvenile hypoparathyroid.

Acquired juvenile hypoparathyroid.

Acquired adult hypoparathyroidism.

Syndromes with diabetes mellitus

Insulin-dependent diabetes mellitus in adolescents.

Non-insulin-dependent diabetes mellitus in adolescents.

Non-insulin-dependent diabetes mellitus in adults.

Diabetes mellitus associated with other syndromes:

DIDMOAD.

Prader-Willi.

Miescher I.

Wiedemann II.

Seip-Lawrence.

Lipoatrophic diabetes mellitus.

Kearns.

Achard-Thiers.

Almström-Hallgren.

Bard-Pick.

Forestier.

Friedreich I.

Herrmann-Aguilar-Sacks.

Heycock-Wilson.

Houssay.

Huntingtong. Lundbaek.

Hemoglobinopathic methemoglobinemia.

Morgagni.

Oppenheim-Urbach.

Chronic porphyria.

Sanctis-Cacchione.

Troell-Junet.

Troisier-Hanot-Chauffard.

Werner.

Flyinn-Aird.

Barjon-Lestrade-Labauge.

Mauriac.

Angiopathic diabetes.

Kimmelstiel-Wilson.

ALLEN-MASTERS SYNDROME

Synonymy

S. of rupture of the broad ligament.

Syndromography

Clínic

This syndrome usually occurs in multiparous women. It is characterized by pain in the pelvic region, pain during sexual relations, dysmenorrhea and back pain.

Physical examination. The cervix can be mobilized in all directions without internal movement.

Paraclinical examinations

Laparoscopy. A certain diagnosis can be made quickly by this method, in which the lesion that causes this syndrome is evident.

Syndromogenesis and etiology

It is due to the rupture of the broad ligament, with angiomatous-looking formations at that level.

Sometimes the etiology is unknown. But it can occur during surgical trauma or an abortion in an advanced period of gestation.

Literature

Allen, W.M. and W.H. Masters: "Traumatic laceration of uterine support, the clinical syndrome and the operative treatment."
Amer. J. Obstet. Gynec., 70:500-5 13, 1955.

Bret, A.J.; M. Bardiaux;J. Brus, et al.: "Algies pelviennes d'origine genitale:; La pelviperitonite chronique sclero-inflammatoire. Etude clinique et histopathologique de 25 cas. A propos du syndrome d'Allen et Masters". Sem. Hop. Paris, 43:173- -182, 1967.

ANGIECTID SYNDROME

Syndromography

Clínic

This syndrome is characterized by the appearance of well-defined, small, painful areas, with local heat, on the back of both legs of the patient during the first month of pregnancy.

Physical examination. Vascular lesions in the form of clusters that are prominent on the skin of the posterior surface of the legs, located intradermally.

Syndromogenesis and etiology

It is proposed that this syndrome is related to the level of circulating female hormones. The lesions are more painful when there is a low level of estrogen or pregnadiol.

UTERINE STROKE SYNDROME

Synonymy

S. of hemorrhagic necrosis of the senile endometrium.

Syndromography

Clinic

This syndrome is characterized by vaginal bleeding, symptoms of heart failure: cough, dyspnea, chest pain and edema in the lower limbs. Hematemesis and melena.

Physical examination. Signs of heart failure, congestive hepatomegaly, tachycardia, cardiomegaly, edema in the lower limbs, postmenopausal vaginal bleeding with integrity of the genital organs.

Paraclinical examinations

Uterine curettage. It allows us to rule out probable uterine carcinoma.

Syndromogenesis and etiology

It is caused by necrosis of the endometrium due to generalized and severe arteriosclerosis of the uterus.

Literature

Novak, E.R. and J. D. Woodruff: Gynecologic and Obstetric Pathology with clinical and Endocrine Relations. ed. 6, Philadelphia, Saunders, 1967.

Daly, J.J. and K. Balogh, Jr.: "Hemorrhagic necrosis of the senile endometrium (apoplexia uteri"). Relation to superficial hemorrhage necrosis of the bowel", New Eng. J. Med.. 278:709-711, 1968.

ASHERMAN SYNDROME

Synonymy

S. of uterine traumatic adhesion.

Syndromography

Clínic

Amenorrhea, recurrent abortions, hypomenorrhea, decreased tion of fertility.

Physical examination. Occluded cervix. Impossibility of passing the explorer to the uterus due to occlusion of the neck canal.

Paraclinical examinations

Difficulty or impossibility of exploring the canal of the uterine cervix due to adhesions at that level.

Hysterography. Defects are fully confirmed by adhesions within the uterine cavity that are not erased with high amounts of contrast medium.

Syndromogenesis and etiology

This condition may occur when curettage is performed on a puerperal uterus, or by repeated curettage, manual removal of the placenta, or intraluminal therapeutic radiation.

Literature

Asherman, J.G.: *Amenorrhea traumatica (Atretica)". J. Obstet. Gynaec. Brit. Emp., 25-30, 1948.
Asherman, J.G.: "Traumatic intrauterine adhesions." J. Obstet. Gynaec. Brit. Emp., 57:892-896, 1950.

PELVIAN CONGESTION SYNDROME

Synonymy

S. Taylor

Syndromography

Clinic

This syndrome affects women of childbearing age. Symptoms occur especially in the premenstrual period.

There is frequently menometrorrhagia, diffuse and poorly defined pelvic pain, deep and sometimes lumbosacral in location. It is accompanied by emotional lability and instability.

Physical examination. On vaginal examination, the cervix appears soft with a large amount of mucous secretion. The uterus is enlarged and there is exquisite pain upon movement, which constitutes an almost pathognomonic sign. There is congestion in the breasts with slight pain on palpation and varicose veins in the lower limbs, frequently.

Paraclinical examinations

Rectal and vaginal touch. The findings mentioned in the physical examination are appreciated.

Laparoscopy. Dilated, tortuous and thickened pelvic veins are observed. The uterus is almost always in retroversion.

Syndromogenesis and etiology

It has been suggested that this syndrome is due to stress of psychogenic origin in women with a history of childhood with family insecurity, as well as signs of immaturity and dependency.

It can also be the result of an excessive hormonal effect on the vessels of the reproductive organs, which would cause engorgement and edema of the pelvic viscera.

Bibliography

Kaufmann, E. (1884) quoted by H.C. Sterus and V.D. Sneeden: "Observations on the clinical and pathological aspects of the pelvic conditions syndrome". Amer. J. Obstet. Gynec., 94:718-732, 1966.
Taylor, H.C., Jr.: "Vascular congestion and hyperemia. Their effect on structure and function in the female reproductive system." Amer. J. Obstet., 57:211-230, 637-653, 654-668, 1949.

CHORIOCARCINOMA SYNDROME

Syndromography

Clinic

This syndrome is consecutive to the hydatidiform mole, and is characterized by abortions, uterine hemorrhage in the second trimester, three of pregnancy. Frequently the first symptoms and signs are expressions of metastases in the lungs, liver, brain or other organs.

Paraclinical examinations

Histology of the uterine mucosa.

Chest radiology. It allows detecting metastases.

Ultrasound. It is especially important in the upper abdomen to try to detect liver metastasis.

Syndromogenesis and etiology

The etiology is unknown.

PLACENTAL DEtachment SYNDROME

Synonymy

S. of uteroplacental apoplexy. Couvelaire's premature placental separation.

Syndromography

Clinic

In this syndrome, vaginal bleeding, localized pain in the pelvis, asthenia and shock occur suddenly.

Physical examination. Rapid increase in the uterus with a sensation of increased texture which gives the impression of firmness.

From the fetal point of view, there is an increase in fetal movements and alterations in the fetal heartbeat: tachycardia and changes in heart sounds.

Paraclinical examinations

Blood. The coagulation study shows hypofibrinogenemia. Hematological alterations that demonstrate renal failure: uremia, electrolyte alterations, etc.

Syndromogenesis and etiology

The symptoms are due to placental abruption that always occurs after 28 weeks and is of unknown cause. The detachment can be partial or total or even affect only the placental edge (rupture of the marginal sinus or hemorrhage of the marginal sinus).

Hemorrhage from the placental surface can dissect beneath the membranes and find its way to the outside, causing external hemorrhage; or it may become trapped and cause occult bleeding, or both.

The following predisposing factors are cited: toxemia, hypertension, chronic glomerulonephritis, trauma, chronic vascular diseases, diabetes and chronic endometritis.

Literature

De Lee, J.B.: “A case of fatal hemorrhagic diathesis, with premature detachment of the placenta." Amer. J. Obstet., 44:785-792, 1901.
Schneider, C.L.: "Defibrination syndrome." Clin. Obstet. Gynec. 7:339-348, 1964.

ECTOPIC PREGNANCY SYNDROME

Synonymy

S. of extrauterine pregnancy.

Syndromography

Clinic

This syndrome has an incidence of 1:300 pregnancies. Straining during defecation, intercourse, and bimanual pelvic examination are cited as factors that precipitate tubal pregnancy rupture. The most common symptoms are irregular vaginal bleeding, absence of menstruation, pelvic discomfort; A few days later hemorrhage begins in the abdominal cavity.

The pain is almost always present and is dull and permanent, radiating towards the back of the trunk. These symptoms are caused by intraperitoneal hemorrhage that can be accompanied by shock when it is massive.

Physical examination. During the gynecological examination, a mass or tumor can be palpated in one of the adnexa. There are signs of peritoneal irritation and sometimes a certain periumbilical bluish discoloration can be seen (Cullen's sign).

Paraclinical examinations

Abdominal puncture. It can be performed at the level of the lower quadrants of the abdomen.

Puncture of the pouch of Douglas. Sometimes it allows obtaining liquid blood as in abdominal puncture.

It is good to note that these methods often fail or confuse.

Ultrasound. It is useful when it specifies the existence of an adnexal tumor mass, but it is often confusing.

Emergency laparoscopy. It is the most important examination and allows the diagnosis to be made with certainty. It also determines the state of the condition and the severity of the complications (ectopic pregnancy, fissured rupture, intensity of bleeding, concomitant injuries). At present, it allows treatment by laparoscopic surgery.

Syndromogenesis and etiology

Chronic inflammatory diseases of the tubes that do not allow the free transit of the fertilized egg to the uterine cavity have been pointed out as possible causes of this syndrome, such as chronic salpingitis, gonorrhea, genital tuberculosis, endometriosis and congenital factors. Chlamydial infection is currently considered the most important cause.

AMNIOTIC FLUID EMBOLISM SYNDROME

Syndromography

Clinic

This syndrome presents with sudden dyspnea, shock, severe hemorrhages, usually in multiparas over 30 years of age with a history of having suffered laborious labors where pitocin was administered.

Physical examination. Tachypnea, dullness on pulmonary percussion, cyanosis, hypotension, uterine relaxation due to profuse vaginal bleeding.

Paraclinical examinations

Blood. Hypofibrinogenemia, prolongation of clotting time.

Syndromogenesis and etiology

This syndrome is due to an embolism of amniotic fluid within the cardiovascular system, which causes chest pain, dyspnea and other symptoms, in addition the decrease in fibrinogen explains the uncontrollable bleeding that accompanies the clinical picture.

Literature

Aguillon, A.; A. Andjus; A. Grayson, et al.: "Amniotic fluid embolism: A review". Obstet. Gynec. Survey, 17:619036, 1962.

Beller, F.K.; G.W. Douglas; C.H. Debrovner, et al.: "The fibrinolytic system in amniotic fluid embolism." Amer. J. Obstet. Gynec., 87:48-55, 1963.

ENDOMETRIOSIS SYNDROME

Syndromography

Clinic

Endometriosis occurs in middle-aged women (third decade) and according to some authors it is more common in women of high socioeconomic status. Some phenotypic characteristics of these patients are cited, such as: thin, anxious, intelligent, perfectionist women who have deferred their offspring for a long time. The condition begins after menarche and disappears with menopause. It is rarely seen in women who have anovulatory cycles, and is more common in those who have menstrual irregularities. Symptoms improve during pregnancy and when these are frequent they can prevent injuries.

Other times it is associated with infertility, progressive and severe pain in the suprapubic region before or during menstruation, dyspareunia, painful defecation, premenstrual spots and hypermenorrhea; Sometimes there may be dysuria and/or hematuria.

Paraclinical examinations

Laparoscopy. It is the most accurate diagnostic method and allows visualization of endometriotic tissue with its typical reddish-brown color, the most frequent location of which is in the pelvic cavity (but also in other abdominal organs). The most important thing is that this exam establishes the differential diagnosis, ruling out pelvic inflammation and tumor or ovarian diseases.

Syndromogenesis and etiology

There are many theories to explain this syndrome. Sampson postulates that fragments of endometrial tissue may be regurgitated with menstrual blood into the peritoneal cavity with subsequent implantation therein. Meller believes that the mesothelial tissue of the peritoneum can evolve and become endometrial tissue. Other authors propose that an endometrial stroma with glandular-looking spaces can form from organized fibrinopurulent exudates.

The etiology is unknown.

DEAD FETUS SYNDROME

Synonymy

Chronic fibrinogen-fibrin conversion syndrome. S. of macerated dead newborn.

Syndromography

Clinic

The diagnosis of this syndrome is based on the clinical manifestations of hemorrhage and fibrinogen disorders, in patients in whom it is known or suspected that they have a dead fetus in the uterine cavity.

The term missed abortion is applied when fetal death occurs before the end of the 20th week of gestation, with retention of the fetus. Although rare, the fetus can occasionally remain intrauterine for months and years and calcify.

There are three degrees of intensity in this syndrome:

1. Potential. No bleeding, fibrinogen below 150 mg% with or without evidence of fibrinolysis.
2. Hidden. No bleeding, fibrinogen below 90 mg%, or high fibrinolytic activity.
3. Obviously. Profuse hemorrhages due to coagulation deficiency with hypofibrinogenemia and hyperfibrinolysis.

Paraclinical examinations

Blood. Serial fibrinogen determinations.

Ultrasound. Evidence of fetal death on ultrasound due to the absence of movement and heartbeat of the fetus.

Syndromogenesis and etiology

It is due to fetal death, with maternal absorption of pregnancy products (amniotic fluid and fetal autolysis products) that produce the activation of fibrinolysis mechanisms, which causes slow defibrination of the blood.

Literature

Weiner, A.E.; OF. Reid; DC Roby, et al.: *Coagulation defects with intrauterine death from Rh sensitization". Amer. J. Obstet. Gynec., 60:1015-1022, 1950.

Hodgkinson, C.P.: R.J. Thompson and A.A. Hodari: "Dead fetus syndrome." Clin. Obstet. Gynec., 7 No. 2:349-360, 1964.

GRAVITATIONAL LEG SYNDROME

Syndromography

Clínica

This syndrome is four times more common in women than in men and is characterized by burning, itching, pain and cramps in the legs and sometimes ulcerations.

Physical examination. There is dilation and tortuosity of the superficial veins of the legs, chronic edema, ulcerations, indurations and dark pigmentation of the skin with stasis and dermatitis.

Syndromogenesis and etiology

It is considered that there is an incompetence of the venous valves that is inherited as a family trait or may be produced by post-phlebitic lesions that cause increased venous pressure. This gives rise to the engorgement of the veins in the lower limbs and the rest of the symptoms as a consequence of stasis.

The etiology is unknown.

Literature

Bauer, G.: “Pathophysiology and treatment of the lower leg stasis syndrome". Angiology, 1:1-8, 1950.

PLACENTARY HEMANGIOMA SYNDROME

Syndromography

Clinic

It is characterized by placental hemangioma, hydramnios, fetal prematurity. Various congenital fetal anomalies.

Paraclinical examinations

Ultrasound. It shows an increase in the fluid of the gestational sac and the hemangioma that detaches a part of the placenta is frequently detected.

Syndromogenesis and etiology

It is considered that the placental hemangioma is what begins the chain of events that determine alterations in the normal circulatory physiology of the fetus.

POSTABORTION FUNCTIONAL HEMORRHAGE SYNDROME

Synonymy

S. of post-abortion metropathy.

Syndromography

Clinic

In this syndrome, vaginal bleeding occurs in women in the reproductive period after a clinical abortion or suspected abortion.

Physical examination. Recent pregnancy, enlarged uterus, soft cervix and vaginal clots.

Paraclinical examinations

Determination of gonadotropins. In blood and urine it helps us diagnose.

Syndromogenesis and etiology

This syndrome is due to the persistence of chorionic villi with a variable degree of viability, which can be identified by histology.

The slow degree of regression of the corpus luteum maintains an estrogen-progesterone imbalance that creates a disorder in the menstrual rhythm, with endometrial hyperplasia.

Literature

Aschoff, L.: Treatise on pathological anatomy. Barcelona, Labor, v. II 1934, p. 927.

Botella-Llusia, J. ;F. Nogales-Ortiz; H. Martínez et al.: “Postabortive functional metrohagia, a clinical and histopathologic study.” Amer. J. Obstet. Gynec., 100-987-996, 1968.

GESTATIONAL HERPES SYNDROME

Synonymy

S. of gestational dermatitis. S. of bullous circinate herpes. S. of gestational prurigo.

Syndromography

Clínica

This syndrome is seen in a ratio of less than 1:5000 pregnancies. It presents during the second trimester with manifestations of severe lesions with redness in the form of a burn accompanied by itching on the face, arms, legs and trunk.

Physical examination. Erythematous, vesicular, bullous, pustular lesions, spread centripetally on the body, starting around the umbilicus.

Paraclinical examinations

Blood. Eosinophilia in more than 30%.

Syndromogenesis and etiology

Gestational herpes is considered a variant of dermatitis herpetiformis, specifically related to pregnancy and the hormonal cycle. It is suggested that it could be due to a high level of circulating gonadotropin.

Literature

Milton. J.L.: The Pathology and Treatment of Disease of the Skin. London, Hardwicke, 1872, p. 205.
Downing, J.G. and O.F. Jilson: "Herpes gestacionis." New Eng. J. Med., 241:906, 1949.

HYPEREMESS GRAVIDICA SYNDROME

Syndromography

Clinic

Hyperemesis gravidarum begins as a simple vomiting, around the sixth week, which gradually increases, despite the treatment imposed. Patients do not even retain ingested liquids. It is accompanied by nervousness, insomnia, and in severe cases it leads to polyneuritis and coma.

Physical examination. Dehydration, weight loss and wasting.

Paraclinical examinations

Urine. Concentrated, albuminuria.

Blood. Electrolyte imbalance. Sometimes hyperbilirubinemia.

Syndromogenesis and etiology

Metabolic changes are invoked, but psychoneurotic factors play an important role.

The etiology is unknown.

SUPINE HYPOTENSION SYNDROME

Synonymy

S. of the inferior vena cava.

Syndromography

Clinic

It occurs in 3-11% of pregnant women in the last month of pregnancy.

Physical examination. Tachycardia. Drop in blood pressure above 30 mm Hg. In few cases, premature placental abruption may occur due to the sudden increase in venous pressure that may be caused by lying in the supine position or by compression of the inferior vena cava.

Syndromogenesis and etiology

This syndrome is produced by compression of the inferior vena cava caused by the occupied uterus, especially at the end of pregnancy, which reduces venous return to the heart and decreases the output volume of this organ.

Literature

Runge, H.: “Venous pressure during pregnancy, delivery and puerperium.” Arch. f. Gynak., 122:142-57, 1924.
Robbins, R.A.; T. Estrara and C. Russe: "Supine hypotensive syndrome and abruptio placenta." Amer. J. Obstet. Gynec., 80:1207-1208, 1960.

PUERPERAL INFECTION SYNDROME

Syndromography

Clinic

This syndrome is characterized by high fever with chills and tachycardia in a newborn. This clinical condition lasts for more than 2 consecutive days, excluding the first 24 hours.

It is accompanied by pain in the pelvic region. Inflammatory lesions may occur in the lungs and septic complications may occur in distant organs.

Physical examination. A uterus is found that has not undergone the normal postpartum involution process.

Paraclinical examinations

Blood. Leukogram with elevated leukocytosis.

Bacteriological examination of the uterovaginal secretion where the pathogenic germ that causes sepsis is isolated.

Chest radiology. It allows detecting inflammatory lung lesions.

Urine. It generally detects urinary sepsis.

Syndromogenesis and etiology

This syndrome is due to the introduction of bacterial sepsis caused by repeated vaginal examinations during laborious labor where premature rupture of the membrane has occurred. Sepsis can be local and limited to the site of placental implantation, extend to the entire endometrium or even affect different locations of the body, causing:

pelvic and femoral thrombophlebitis, pulmonary infarction, abscesses.

PLACENTARY INSUFFICIENCY SYNDROME

Synonymy

S. of Ballantyne. S. de Runge, S. de Clifford. S. of small fetus. S. of prolonged gestation. Post-maturation S. S. of placental dysfunction.

Syndromography

Clinic

Decreased fetal movements after 30 weeks of pregnancy.

According to the post-maturity time of the fetus during its intrauterine life, three degrees have been described:

Grade I. Peculiar habit, they are long, thin, with skin that is large and makes folds on the arms and buttocks (collodion cutis) with extensive peeling on the hands, feet and abdomen, without vernix caseosa; long nails and thick hair.
"Awake" looking child with open eyes (little old man).

Grade II. The same as the previous one but meconiorrhea occurred and the skin and cord were tinged green at birth.

Grade III. The fetus suffered fetal distress and was born with yellow staining of the cord and nails.

Some authors consider that this syndrome and the so-called post-maturity syndrome have certain characteristics that allow them to be differentiated, but in a certain way they all present similar clinical pictures.

Physical examination. Delayed uterine growth after 30 weeks of pregnancy, the fetus is small even though it is full term. Intrauterine fetal death occurs with some frequency.

Paraclinical examinations

Ultrasound. It allows determining the fetal head size.

Amniocentesis. 1, If the liquid is clear, do not intervene.

2, Monitor the proportion of estriol in the pregnant woman's urine twice a week; if it exceeds 25 mg/24 h there is no risk.
3, If amniotic fluid is not obtained, test for oxytocin and if negative, monitor estriol. 4, If there is meconiorrhea, induce labor.

Urine. Decreased excretion of estriol. Increase in atropine transfer time.

Syndromogenesis and etiology

This syndrome is due to a decrease in placental vascular flow with an increase in fibrosis of the placental tissue and a reduction in the size of the placenta with a weight of less than 200 g.

Placental aging causes dysfunction with fetal hypoxia and hyponutrition, hemoconcentration and hypoglycemia. In 40% of cases there are placental anomalies, meconium staining and degenerative disorders; in some, meconia aspiration and pneumonia; in 15%, infections.

It is estimated that some clinically recognizable form of placental dysfunction occurs in 12% of all births.

Frequency, according to gestational age: more than 43 weeks, 5%: more than 42, 10% and more than 41, 25%.

The etiology can be varied: persistence of the fetal head above the pelvic inlet, congenital malformations and genital infantilism in mothers.

Literature

Ballantyne, J.W.: The problem of the post mature infant." J. Obstet. Gyn. Brit. Emp., 2:521-554, 1902.

Clifford, S.H.: "Postmaturity." Advanc. Pediat., 9:13, 1939. Grant, K.E. et al,: in Avery: Neonatology. Philadelphia, Lippincott, 1975. p. 58.

Rumbolz, W.L.; MC Edwards and L.s. McCoogan: "The small full term infant, placental insufficiency." West. J. Surg., 69:53-60, 1961.

Runge, H.: "Die Langdaurnde Schwangerschaft." Zlbl. Gynaek. 63:972. 1939.

RING NECK RING LESION SYNDROME

Synonymy

S. of traumatic separation of the uterine cervix.

Syndromography

Clinic

This syndrome is characterized by abundant and sustained bleeding after childbirth.

Physical examination. The ring-shaped neck injury caused by the child's head can be visualized. Frequently, upon inspection, the cervix appears hanging by a flap of tissue.

Paraclinical examinations

Colposcopy. It allows you to visualize the uterine cervix and check the lesions that cause the hemorrhage.

Syndromogenesis and etiology

This syndrome is due to factors that prolong labor; cervical dystocia (more common in primiparas), cephalopelvic disproportion.

Literature

Fliegner, J.R.: Sponatenous annular detachment of the cervix'". Med. J. Aust., 1:438-441, 1968.

Ingraham, C.B. and E.S. Taylor: *Spontaneous annular detachment of the cervix during labor". Amer. J. Obst. Gynec., 53:873-877, 1947.

MAYER--ROKITANSKY-KUSTER--AUSER SYNDROME

Synonymy

S. de Rokitansky-Kuster-Hauser. Vaginal uterine anomaly.
S. of uterus bipartitus solidus rudimentarius cum vagina solida.
Rokitansky S.

Syndromography

Clinic

Second cause in order of frequency of primary amenorrhea, after gonadal dysgenesis.

One in every 100,000 women is born without a vagina. Most patients are discovered after puberty is expected in mid-adolescence. The patient consults to find out why she does not have menstruation if she is already physically a woman with all the secondary sexual characteristics.

Sometimes there is abdominal pain, cyclical due to retrograde menstruation or hematometra.

Mental retardation is exceptional.

Physical examination. Normal height and intelligence; breasts, axillary and pubic hair and external habit of feminine type; The uterus can vary from almost normal, without the duct to the introject, to the more characteristic rudimentary bicornuate cords. Hypoplasia or absence of vagina. Impossible to perform vaginal touch despite the existence of a vulva.

In 90% of cases there is no uterus or it is bifid.

Paraclinical examinations

Chromosome study. Karyotype 46XX.

Ovulatory plasma LH maxima and biphasic temperature curves during the cycle suggest normal ovarian function.

Ultrasound. Presence of ovaries and almost always absence of uterus.

Pneumogynecography. It is more demonstrative in these cases.

Laparoscopy. It is the most complete examination and also allows us to specify not only the size of the ovaries but also their functionality.

Ultrasound. Rudimentary, bicornuate uterus, with persistence of tubes and ovaries. Renal agenesis or hypoplasia in almost half of the cases.

Radiology. Vertebral anomalies may be seen and/or sacks.

Descending urography. In detection of anomalies finals.

Audiometry. Conduction deafness due to malformations of the ossicles of the ear.

Syndromogenesis and etiology

Autosomal dominant mutation limited to sex in familial cases. Most are sporadic cases. It is not known if these represent new mutations or if they have a multifactorial cause. In familial cases, there is frequently variable expressivity: some members have only skeletal or renal anomalies, others have anomalies of the Müllerian derivatives such as a double uterus. Bilateral renal aplasia is often related to the absence of the uterus and vagina. Klippel-Feil syndrome frequently coexists.

It is considered that it could be a virus that affects the development of the Müllerian system around 54 days after fertilization, which causes two rudimentary uterus outlines to remain and no vagina to form.

The ovaries come from other structures and are almost never affected.

The external genitalia derived from the urogenital sinus are normal, as are the secondary sexual characteristics.

With surgical repair and formation of an artificial vagina, a woman can have normal sexual relations and orgasm. She is just unable to conceive.

MEIGS SYNDROME

Synonymy

S. of pleural effusion-ascites-ovarian. S. de Demons- - Meigs.

Syndromography

Clinic

This syndrome is characterized by chest pain, shortness of breath, urinary incontinence.

Physical examination. Ascites, hydrothorax (62% on the left side, 11% on the right side, 24% bilateral), adnexal tumor, uterine prolapse, edema in lower limbs infrequent.

Paraclinical examinations

Pleural and abdominal punctures. Extraction of light yellow fluid (transudate) sometimes somewhat darker.

Chest radiology. Image of pleural effusion.

Abdominal ultrasound. Presence of ascites.

Laparoscopy. Solid-looking ovarian tumor.

Syndromogenesis and etiology

Solid ovarian tumor, generally benign, fibroma, granulosa cell tumor or Brenner tumor, thecoma. The mechanism of formation of ascites and hydrothorax is unknown.

Removal of the tumor makes liquid spills disappear.

Literature

Meigs, J.V. andJ.W. Cass: Fibroma of the ovary with ascites and hydrothorax, with a report of 7 cases." Am. J. Obstet. Gynecology, 33:249-267, 1937.

MENDELSON SYNDROME

Synonymy

S. obstetric bronchopulmonary aspiration. S. acid pulmonary aspiration. S. of aspiration peptic pneumonia.

Syndromography

Clinic

This syndrome is characterized by tachypnea, tachycardia, noisy breathing and sudden onset cyanosis.

Physical examination. On pulmonary auscultation, hoarse and wheezing rales are heard. There is tachycardia and if the condition progresses, it leads the patient to collapse with arterial hypotension and shock. Pulmonary edema may occur.

Paraclinical examinations

Blood. Fall in arterial oxygen tension.

Radiology. Irregular or confluent radiopacities of bilateral location.

Syndromogenesis and etiology

It is caused by the aspiration of gastric contents with acid-peptic secretion during anesthesia in pregnant patients, which causes the brochopulmonary reaction and circulatory symptoms.

Literature

Dines, D.E., et al.: "Aspiration pneumonitis: Mendelson's syndrome." J.A.M.A., 176:229. 1961.

Mendelson, C.L.: The aspiration of stomach contents into the lung during obstetrics anesthesia." Am. J. Obstet. Gynecol., 52:191, 1946.

Richman, H., et al.: "Mendelson's syndrome." Am. J. Surg., 120:531, 1970.

MENOPAUSAL SYNDROME

Syndromography

Clinic

Suffered by some women during the climacteric. Facial flushing, chills, sweating, tachycardia, palpitations, nervousness, irritability, depression, headache, loss or increase in libido, metrorrhagia, vulvar pruritus, atrophic vaginitis, painful or hypertrophic breasts, bursitis, bone pain.

Paraclinical examinations

Blood. Increased FSH, increased cholesterol.

Radiology. Osteoporosis, arthritic changes.

Syndromogenesis and etiology

Declining ovarian function with deficiency in estrogen secretion.

Literature

Paschkis, K.E.; A.E. Rakoff; A. Cantarow, et al.: Clinical Endocrinology. ed.3, New York, Harper-Row, 1967.

MICROMIOMAS SYNDROME OF THE UTERUS

Syndromography

Clinic

This syndrome is characterized by menorrhagia that does not respond to the administration of hormonal therapy, curettage or oxytocics. Menstrual regulation time is normal.

Physical examination. On recto-vaginal examination, the uterus is palpably slightly enlarged, firm in consistency, without irregular nodules on its surface.

Paraclinical examinations

Uterine curettage and endometrial biopsy. It reveals muscular hypertrophy reminiscent of a diffuse fibroid-type miliary process with an increase in the number and tortuosity of the vessels and malignancy of the wall.

Syndromogenesis and etiology

Micromyomas interfere with the contractility of the uterine muscular layer, which explains the hemorrhage.

The etiology is unknown.

Bibliography

Hiller, R.I.: “Mycromyomas of the uterus with severe menorrhagia: a syndrome". Amer. J. Obstet. Gynec., 87:163-165, 1963.

HYDATIFORM MOLE SYNDROME

Syndromography

Clínic

The incidence of this syndrome is 1 in every 2,000 pregnancies and is more common in the Far East. Bleeding in the form of spots or profuse, nausea and vomiting. Pain due to rapid growth of the uterus.

Physical examination. Uterine growth beyond the age of pregnancy. Bilateral growth of the adnexa, due to ovarian luteal cystic formations. High blood pressure during the second trimester.

Paraclinical examinations

Blood. Increased chorionic gonadotropin.

Urine and organic fluids. Increased gonatropins.

Syndromogenesis and etiology

The etiology is unknown.

PLACENTA PREVIA SYNDROME

Syndromography

Clínic

Placenta previa occurs in 1:100-1:200 cases and is more common in multiparas. There is bleeding with little pain during the third trimester of pregnancy. Hemorrhage can occur without prior warning and without being accompanied by preeclampsia.

Physical examination. Vaginal examination, under sterile conditions, reveals the presence of the placenta in the lower segment of the uterus, partially or completely covering the uterine orifice of the cervix.

Paraclinical examinations

Placentography. Cone without contrast can be performed in the bladder or rectum using 12.5% sodium iodide. Serum albumin labeled with radioactive IV iodinate that localizes the placenta can also be used.

Syndromogenesis and etiology

Little is known about the causes of placenta previa.

It is suggested that it could be related to a vascularization defect of the decidua, as a result of an inflammatory or atrophic process.

PREMENSTRUAL SYNDROME

Syndromography

Clinic

In this syndrome, physical and psychological or behavioral symptoms are associated, which begin in the two weeks before menstruation, decrease during it and fade a few days after it begins.

Among the most common symptoms are: depression, anxiety, mastodynia and food "cravings." Emotional instability, dizziness, constipation or diarrhea, edema in the lower limbs, mastodynia and pain in the hypogastrium, migraine or headache.

In addition, the patient's conditions may be exacerbated: acne, asthma, epilepsy.

It is believed that 70-90% of women of reproductive age present recurrent premenstrual symptoms, with greater or lesser intensity, and 20-40% of them suffer some mental or physical incapacity.

The symptoms tend to disappear as the patient ages 30 or 40.

The following have been identified as predisposing factors: multiparity, history of preeclampsia, alcohol and/or narcotic abuse, and affective disorders.

Syndromogenesis and etiology

Most researchers consider that it is a physiological alteration, exacerbated by environmental and personality factors (stress, patient's attitude towards menstruation, reaction of the spouse and/or family). The following hypotheses have also been considered: deficiency of progesterone, betaendorphins, vitamin B6 or magnesium.

Others attribute it to excess prostaglandins, prolactin, estrogens or corticotropin.

It is essential to rule out organic conditions that could simulate this syndrome, such as: uterine fibroid, pelvic inflammatory disease, endometriosis, dysmenorrhea, etc.

Literature

O'Brien, PM.: "The premenstrual syndrome: a review" J. Reprod. Med.,: 30 (2):113-126, 1985

Youngs, D.D.: “Assessing premenstrual syndrome in a private practice setting.” Postgrad. Obstet. Gynecol. 6(19):1-6, 1986.

PSEUDO-MEIGS SYNDROME

Syndromography

Clinic

The clinical picture is the same as that of Meigs syndrome, except that the location of the tumor may be located in the ovary, tubes, uterus, or in the round ligament.

After tumor excision, ascites and hydrothorax should not necessarily disappear as in the case of Meigs syndrome. The final result depends on the histological nature of the tumor and the presence or absence of metastasis in malignant cases.

Paraclinical examinations

Radiology. Shows pleural effusion.

Laparoscopy. It allows diagnosing the gynecological lesion and checking the ascites, its intensity and characteristics. Ovarian tumor.

Syndromogenesis and etiology

The etiology is unknown since the cause of the ascites or hydrothorax could not be determined.

Literature

Meigs, J.V.: "Pelvic tumors other than fibromas of the ovary with ascites and hydrothorax." Obstet. Gynec., 3:471-486, 1954.

Brenner, W.E. and R.B. Scott: Meig's-like syndrome secondary to krukenberg's tumor." Obstet. Gynec., 31:40-44, 1968.

RACINE SYNDROME

Synonymy

Premenstrual salivary S.

Syndromography

Clinic

Tumor of one or more salivary glands, which occurs 4-5 days before menstruation and disappears with the beginning of it.

Paraclinical examinations

Sialography. Edema and transient dilation of the salivary ducts are found in the salivary glands.

Syndromogenesis and etiology

It is considered to be part of premenstrual tension syndrome.

Literature

Racine, W.: Le syndrome salivaire prémenstruel". Schweiz. Med. Wschr., 69:1204-1 207, 1939.

DELAYED MENSTRUAL PERIOD SYNDROME

Syndromography

Clinic

This syndrome appears associated with the beginning of sexual life or a change in this pattern. Menstrual delay can last between 10-60 days followed by abnormal uterine bleeding.

Physical examination. Normal anatomical and functional signs.

Paraclinical examinations

Pregnancy tests. Negatives.

Uterine curettage. A proliferative endometrium is found or early secretory.

Puncture of the Douglas cul-de-sac. 5-20 mL of serohematic fluid can be

Syndromogenesis and etiology

This syndrome must be differentiated from incomplete abortion, incipient abortion and extrauterine pregnancy.

The etiology is psychogenic, following the beginning of sexual relations, changes in these patterns or fear of pregnancy.

Literature

Soferman, N. and M. Haimov: Psychosexual menstrual disorder". Harefuah. 64:54-55, 1963.

_____ The syndrome of unexplained delayed menstrual period'. Amer. J. Obstet. Gynec. 91:137-141. 1965.

MATERNAL TOXEMIA SYNDROME

Synonymy

S. of preeclampsia and eclampsia. S. of the EPH complex (edema, proteinuria and hypertension).

Syndromography

Clínica

This syndrome occurs in Rh-negative pregnant women around 28-30 weeks through an isoimmunization process.

Physical examination. Weight gain, refractory edema (E) to commonly used diuretics, small elevation in blood pressure (H). These symptoms and signs put isoimmunized mothers at risk of intrauterine fetal death.

Sometimes seizures may occur.

Paraclinical examinations

Blood. Prior determination of the Rh factor in all pregnant women.

Urine. Proteinuria (P).

Syndromogenesis and etiology

A possible disorder of aldosterone secretion is proposed that would give rise to fluid retention in the fetus and the mother, with a great increase in the contents of the uterus, which could produce anoxia of the decidua and perhaps cause an increase in the production of hysterotonin and hence the symptoms of toxemia. There is a certain familial tendency to develop preeclampsia.

The etiology is unknown.

Literature

Jeffcoate, TN.A. and J.S. Scott: "Some observations on the placental factor in pregnancy toxemia." Amer. J. Obstet. Gynec.., 77:475-489, 1959.

Nicolay, K.S. and H.L. Gainey: "Pseudotoxemic state associated with severe Rh isoimmunization." Amer. J. Obtest. Gynec... 89:41-45, 1964.

INTRAUTERINE TRANSFUSION SYNDROME

FROM TWIN TO TWIN

Synonymy

S. of arteriovenous transfusion in newborns.

Syndromography

Clinic

In this syndrome, one of the twins (recipient) is larger, presents polycythemia and hypervolemia, enlargement of the heart with myocardial hyperplasia. Arterial, systemic and pulmonary hypertension. The other twin (donor) is pale with hypovolemia, decreased size of his viscera;
Therefore there is a decrease in body size between the two twins. The recipient twin is almost always associated with polyhydramnios.

Paraclinical examinations

Ultrasound. If there is hydramnios with twin pregnancy or a large difference in size between the twins, this syndrome should be suspected.

Placentogram with radioisotopes. It is useful.

Syndromogenesis and etiology

This syndrome occurs in monozygotic twins that allows blood transfusion from one fetus to another. The transfusion occurs through the conduits between the two circulations: artery-artery, artery-vein and vein-vein.

The etiology is unknown.

Literature

Driscoll, S.G.: "Multiple gestation." Ped. Clin. N. Amer., 12:498- -502. 1965.
Simopoulas, A.P.: "Arteriovenous transfusion syndrome in newborn twins." GP., 33: 141, 1966.

MATERNAL-FETAL TRANSFUSION SYNDROME

Syndromography

Clinic

The clinical picture is the same as that of neonatal polycythemia syndrome.

Paraclinical examinations

Blood. High hemoglobin and hematocrit values. Finding of maternal erythrocytes in the blood of the newborn.

Presence of beta 2M globulin in the newborn's blood.

Syndromogenesis and etiology

This syndrome is caused by a maternal-fetal transfusion that occurred during pregnancy.

Literature

Hedenstedt, S. and J. Naeslund: "Investigations of the permeability of the placenta with the help of elliptocytes." Acta Med. Scand., 170 (suppl). 126-134, 1946.
Michael, A.F. and A.M. Mauer: “Maternal-fetal transfusion as a cause of plethora in neonatal period." Pediatrics, 28;458- -461, 1961.

VASA PREVIA SYNDROME

Syndromography

Clinic

This syndrome occurs during the third trimester of pregnancy. There is discrete blood loss through the vagina.

Physical examination. Irregularity of the fetal focus during uterine contractions. Vaginal bleeding.

Paraclinical examinations

Vaginal hemorrhagic discharge test. Positive for fetal hemoglobin; However, it is negative in the case of maternal hemorrhage.

Syndromogenesis and etiology

It is due to rupture of one or more vessels of the umbilical insertion, due to the pressure of fetal presentation. In addition, there is rupture of the amniotic sac and velamentous insertion of the umbilical vessels.

Literature

Naftolin, F. and D.R. Mishell, Jr.: "Vasa previa," report of 3 cases." Obstet. Gynecol., 26:561-565, 1965.
Torrey, W.E., Jr.: "Vasa previa." Amer. J. Obstet. Gynec., 63:146-152, 1952.

YOUNG-PAXSON SYNDROME

Synonymy

Paxson's traumatic S. S. obstetric-gynecological- -traumatic.

Syndromography

Clinic

This syndrome occurs in the following obstetric conditions: retroplacental hemorrhage, trauma during labor, uterine rupture, twisted ovarian cyst with blood extravasation. The symptoms are shock followed by anuria, renal failure and later hypertension and signs of uremia.

Paraclinical examinations

Blood. Hyperkalemia, increase in nitrogen catabolites.

Urine. Hematuria, pigmentary and granular casts.

Electrocardiogram. Signs of hyperkalemia.

Syndromogenesis and etiology

The most common etiological causes are: trauma with shock that can cause retroplacental hemorrhage. Other pathological conditions may be uterine rupture and twisted ovarian cysts with blood extravasation.

The fundamental mechanism is prolonged ischemia caused by trauma (as in Bywater syndrome).

Literature

Paxson, NE:LJ. Golub and R.M. Hunter: "The crush syndrome in obstetrics and gynecology." J.A.M.A., 131:500-504, 1946.

Young, J.: "Renal failure after utero-placental damage." Brit. Med. J.. 2:715-718, 1942.

YOUSSEF SYNDROME

Syndromography

Clínica

This syndrome is characterized by cyclic hematuria, absence of vaginal bleeding during menstruation and sometimes urinary incontinence.

Paraclinical examinations

Radiology. Cystography and hysterosalpingography show a utero-cystic communication.

Syndromogenesis and etiology

It is due to a communication between the uterus and the bladder after surgical interventions, in low cesarean sections, for example, with the characteristics that the lesion is of a valvular type, passing blood from the uterus to the bladder, but not to the intestine.

versa.

Literature

Youssef, A.E.: “Menouria following lower segment cesarean section: a syndrome." Am. J. Obstet. Gynecol., 73:759, 1957.

Medeiros, A. de S., et al.: Youssef's syndrome". J. Urol.. 109: 828, 1973.

ACANTHALASIA SYNDROME

Synonymy

S. of Takahara.

Syndromography

Clinic

Common in Japanese and Koreans. It has also been reported in whites.

Half of the patients are asymptomatic. It presents with a severe chronic infection of the mouth with gangrenous lesions that include alveolar destruction.

Paraclinical examinations

Blood. The addition of hydrogen peroxide to the blood of these patients turns it blackish brown due to methemoglobinemia (normal blood maintains its color).

Syndromogenesis and etiology

Genetic catalase deficiency in tissues and cells, including blood, erythrocytes, etc., decreases the mouth's resistance to normal flora.

The catalase (CAT) gene has been located in 1lp. The Japanese type of acathalassemia is due to a G to A substitution splicing mutation in the fifth position of intron 4.

Literature

Takahara., S.: “Progressive oral gangrene probably due to lack of catalase the blood." Lancet, 2:1101-1104, 1952.

Wen, J.K. et al.: Molecular analysis of human acatalasemia identification of a splicing mutation". JMolec. Biol., 211:383-393, 1970.

ADDISON-BIERMER SYNDROME

Synonymy

S. of pernicious anemia. by Biermer.

Syndromography

Clinic

It is a process that affects both sexes. There is weight loss, anorexia, repugnance to certain foods, belching, marked asthenia, yellowish pallor and sometimes subicterus. Glossitis with burning tongue, diarrhea and neurological complications, caused by deep sensitivity disorders.

Paraclinical examinations

Blood. There is macrocytic anemia (megalocytosis), hyperchromia. Increase in mean corpuscular volume. There may be a decrease in platelets. Increased indirect bilirubin.

Elevated serum iron.

Medulogram. Hyperplasia of the erythropoietic system with megaloblastic changes and increased normoblasts.

Other exams. They show: histamino-resistant anachlorhydria. Antibodies against intrinsic factor and parietal cells.

Schilling test. Positive.

Syndromogenesis and etiology

An autoimmune mechanism is proposed, there is gastric atrophy with a deficiency of the intrinsic factor necessary for the absorption of vitamin B12, due to atrophy of the gastric glands.

Literature

Addison, T.: On the constitutional and local effects of the disease of the adrenal capsules. London, 1875.

Biermer, A.: "Form von progressiver pernicioser anamie." Korresp. Bl. Schweiz. Arzt. 2:15-18. 1872.

CONGENITAL AFIBRINOGENEMIA SYNDROME

Synonymy

S. of Rabe-Salomőn. S. of factor I deficiency.

Syndromography

Clinic

It affects both sexes and manifests itself from birth.

There is bleeding from the umbilical cord. This is severe after trauma or in the postoperative period.

Ecchymoses, hematomas, epistaxis, hemoptysis, hematuria occur. Hemorrhage in the central nervous system.

There are asymptomatic periods. Menstruations are normal.

Physical examination. Hemarthrosis or petechiae are rarely present.

Bone pseudocysts may occur.

Paraclinical examinations

Blood. There is absence of fibrinogen. Prolongation of clot retraction. Prothrombin time may be prolonged. Prolonged " bleeding" time. Rest of the normal coagulation factors.

Syndromogenesis and etiology

There is failure in fibrinogen synthesis due to nutations in any of the three fibrinogen genes. It is inherited in an autosomal recessive manner.

Literature

Blomback, M.; B. Blomback; E.FMammen, et al.: "Fibrinogen Detroit-a molecular defect in the N-terminal disulphide knot of human fibrinogen." Nature, 218:134-137, 1968.

Fletcher, A.P.; N. Alkjaersig and S. Sherry: “Pathogenesis of the coagulation defect developing during pathological plasma proteolytic (fibrinogen proteolysis and circulating fibrinogen breakdown products).” J. Clin. Invest., 41:896-916, 1962.
Rabe, F. and E. Salomon: Ueber Faserstoffmangel im Blute bei einem Falle von Haemophile". Deutsch. Ark. Klin. Med, 132:240-244, 1920.

Sharp, A.A.: Pathological fibrinolysis". Brit. Med. Bul., 20:240-245, 1964.

ACUTE AGRANULOCRTOSIS SYNDROME

Synonymy

S. de Schultz. S. of agranulocytic angina. S. of malignant neutropenia. S. of agranulocytic necrotizing mucositis.

Syndromography

Clinic

This syndrome is more common in adult women. With a sudden onset, it is characterized by prostration, fever, chills and polyarthritis, which make up a hyperpyretic septic condition. Ulcerations of the mucous membranes, mainly of the oropharynx, tonsils and mouth, with circumscribed necrosis and formation of diphtheroid membranes. Painful lymphadenopathy.

Paraclinical examinations

Blood. There is leukopenia below 1000, almost always with a complete absence of neutrophils, with toxic granules and changes of the nucleus and cytoplasm. There is no anemia or thrombocytopenia.

Medulogram. It may be normal. Hypoplastic or hyperplastic marrow. In cases with hypoplastic marrow, immature myeloid elements are seen.

Syndromogenesis and etiology

This syndrome may be idiopathic or secondary to ionizing radiation. Chemical agents: pyramidon, sulfas, arsenic, sleeping pills, especially in subjects with idiosyncrasies to drugs.

Literature

Miale, J.B.: Laboratory Medicine, Hematology. ed. 3, St. Louis, Mosby, 1967.

Schultz, W.: "Über eigenartige Halserkrankungen." Deutsch. Med. Wschr., 48:1495-1496, 1922.

FANCONI ANEMIA SYNDROME

Synonymy

S. of familial panmyeloptysis. S. of chronic idiopathic pancytopenia. S. of congenital aplastic anemia. S. of congenital pancytopenia. S. of childhood familial perniziform anemia.

Syndromography

Clínic

This syndrome is more common in men. It is manifested by anemia, fatigue, and paleness that appears within the first 8 years of age. Recurrent infections, easy bleeding, mental and growth retardation occur.

There are brown pigmented spots on the skin and also vitiligo. Intense bone pain, anemia. Peanut-hemorrhagic festivities. Recurrent infections. Infantilism.

Poor hair growth.

Different congenital anomalies are associated: microcephaly, microphthalmia, strabismus, deafness, hypogenitalism. Tendency to malignant lesions especially leukemia in the ratio of 1:12; in the general population it is 1:450.

Paraclinical examinations

Blood. Pancytopenia. Normochromic anemia, macrocytes, target cell. Mild reticulocytosis, generally neutropenia, and nuclear alterations that simulate pseudo Pelger. Immature cells can be found.

Thrombocytopenia. Elevated serum iron. Normal bilimubin.

Normal liver function tests and bilirubin.

Medulogram. At the beginning there is hyperplasia with signs of maturation arrest, later there is progressive hypoplasia and aplasia. Erythrocyte hexokinase deficiency.

Coomb's test. Negative.

Kidney x-ray. Different anomalies are seen: horseshoe kidney, dystopia.

Syndromogenesis and etiology

This syndrome is due to an autosomal recessive genetic defect. It appears to be a defect in DNA separation. A disorder has been suggested in the passage of enzymes related to DNA separation that are produced in the cytoplasm and have to reach the nucleus. It presents chromosomal instability such as Bloom syndrome and ataxia-telangiectasia. Some authors believe that Fanconi anemia and Blackfan-Diamond syndrome are mutations of the same locus.

Literature

Fanconi, G.: "Familiare infantile, pernicioseahnliche Anämie (pernizioses Blutbild und Konstitution)". Jahrb. F. Kinderh., 117:257-280, 1927.

Rohr, K.: “Familial panmyeolophthisis, Fanconi syndrome in adults.” Blood, 4:130-141, 1949.
Von Lohr, G.W. and H.D. Waller: "Beitrag zur Pathogenese der Fanconi Anämie." Blut. 15:321-329, 1967.

DEANEMIA-HEMATURIA SYNDROME

Syndromography

Clínica

Symptoms vary according to the season in which the syndrome occurs.

There are constant symptoms such as: asthenia, fever, tachycardia, anemia, vesicular-erythematous dermatitis, which appear at any season.

In the summer there are: anorexia, vomiting, gastralgia, diarrhea, jaundice, dehydration and nervousness. In winter: edema, oliguria, hematuria, albuminuria, cylindruria and increased anemia.

Syndromogenesis and etiology

The cause is unknown.

Literature

Villar, J.B.: “Epidemic anemic-hematuric syndrome.” Rev. Asoc. Med. Argent., 62:649, 1948: Abst., J.A.M.A., 140:364, 1949.

HEMOLYTIC ANEMIA SYNDROME DUE TO GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY (GOPD)

Synonymy

S. of congenital hemolytic anemia Dacie I. S. of primaquine sensitive hemolytic anemia.

Syndromography

Clínica

This syndrome occurs in men who live in the Mediterranean area, rarely in women (homozygous or only partially expressed in XX heterozygotes).

Various clinical forms can be adopted: spontaneously at any age or induced by exposure to medications such as primaquine and sulfonamides. Hemolytic crisis or more or less compensated chronic hemolysis. History of jaundice at birth. In children there is splenomegaly. In some cases cataracts occur.

Paraclinical examinations

Blood. Anemia, macrocytosis and reticulocytosis. Normal oSmotic fragility. Autohemolysis that is corrected by adding glucose. There is hyperbilirubinemia. Glucose-6- deficiency -phosphate.

Coomb's test. Negative.

Syndromogenesis and etiology

G6PD is the enzyme that participates in the conversion of glucose-6-phosphate to 6-phosphogluconate, it is the only way to obtain NADPH which in turn maintains the concentration of reduced glutathione and this maintains the integrity of the erythrocyte membrane in the presence of oxidizing substances. The G6PD gene has been located on chromosome Xq28; There are many mutations that affect this gene and produce variants of which there are two that cause hemolytic anemia only in the presence of oxidative stress: G6PDA and G6PD Mediterranean or B-.

Individuals with these mutations (A- and B-) of the GoPD gene are asymptomatic under normal conditions, but when exposed to oxidative stress such as exposure to certain drugs (nitrofurantoin, primaquine, sulfacetamide, among others), infections and ingestion of beans, they develop acute hemolytic anemia.

It is inherited following a Mendelian recessive pattern linked to the X chromosome.

According to the level of enzymatic activity, five classes of GOPD variants have been classified:

1. Enzyme deficiency with chronic non-spherocytic hemolytic anemia.
2. Severe enzyme deficiency (G6PD Mediterranean).
3. Moderate enzyme deficiency (G6PDA-).
4. With almost normal enzymatic activity.
5. With increased enzymatic activity.

Literature

Bentler, E.: "Glucose-6-phosphate dehydrogenase deficiency." New. England. J. Med., 324:169-174, 1991.

Cordes, W.: "Experiences with plasmachin in malaria: Preliminary reports." 15th. Annual Report, United Fruit Company (Med. Dept.), 1926, p.66.

Westring, D.W. and A.V. Pisciotta: “Anemia cataracts, and sei zures in patient with glucose-6-phosphate dehydrogenase deficiency." Arch. Int. Med., 118:385-390, 1966.

Wintrobe, M.M.: Clinical Hematology. ed. 6, Philadelphia, Lea & Febiger, 1967.

HEMOLYLIC ANEMIA SYNDROME DUE TO PYRUVATOKINASE DEFICIENCY

Synonymy

S. of pyruvateguinase deficiency. S. of congenital hemolytic anemia. S. of congenital hemolytic anemia type Dacie II.

Syndromography

Clinic

It affects both sexes equally. It settles in childhood.

There is weakness, fatigue. Symptoms are exacerbated when there is an intercurrent illness or during surgery. Jaundice, paleness, splenomegaly; The appearance of hepatomegaly is rare, as are chronic ulcerations of the lower limbs.

There may be cholelithiasis.

Paraclinical examinations

Blood. Anemia. With relative macrocytosis, reticulocytosis and the variable appearance of nucleated erythrocytes. Normal leukocytes and platelets, moderate hyperbilirubinemia. Haptoglobin decreased or absent. Normal osmotic fragility. Positive autohemolysis test, which is not corrected by the addition of glucose and adenosine.

Coomb's test. Negative.

Ferrokinetic study. It reveals a quick replacement of the iron. Shortened red blood cell survival.

Fecal urobilinogen. Increased.

Syndromogenesis and etiology

There is erythrocyte pyruvatokinase deficiency.

It is transmitted by autosomal recessive inheritance with a variable degree of clinical severity between homozygotes and heterogotics.

Literature

Dacie, J.V.; P.L. Mollison; N. Richardson, et al.: "Atypical congenital haemolytic anaemia." Quart. J. Med., 22:79-98, 1953.

Valentine, W.N.; K.R. Tanaka and S. Miwa: "A specific erythrocyte enzyme defect (pyruvate kinase) in three subjects with congenital non-spherocytic hemolytic anemia." Trans. Ass. Physicians, 74:100-110, 1961

HOT ANTIBODY TYPE IDIOPATHIC HEMOLYTIC ANEMIA SYNDROME

Syndromography

Clinic

It occurs at all ages with a slight predominance in women. Sudden or insidious installation. Occasionally there is a history of infections.

Variable symptoms: acute hemolytic crisis, accompanied by fever, sometimes shock. Prostration up to coma or moderate chronic anemia, asthenia and dyspnea on exertion. There is hemoglobinuria, fecal pleiochromia.

Occasionally: superficial thrombophlebitis, gallstones, precordial pain, headache, hemorrhages.

Physical examination. Paleness, jaundice, splenomegaly.

Paraclinical examinations

Blood. Anemia, usually macrocytic. Anisopoikilocytosis, intense, spherocytosis, reticulocytosis, small number of siderocytes, normoblastemia. In the acute periods there is leukocytosis and in the chronic periods there is neutropenia. Numbers of normal or decreased platelets (see S. Evans).

Hyperbilirubinemia. Absence of haptoglobin in acute periods. Increased osmotic fragility, as well as autohemolysis. Shortened red blood cell survival.

Coomb's test. Positive.

Special techniques. To demonstrate special antibodies. Normal serum supplement.

Syndromogenesis and etiology

A fundamental hereditary aberration of the immune system is suggested in cases whose relatives suffer from other types of diseases such as lupus erythematosus, rheumatoid arthritis.

The etiology is unknown.

Literature

Dacie, J. V.: The haemolytic Anaemias. New York, Grune & Stratton, 1963.

ATROMBIA SYNDROME

Synonymy

S. of platelet factor 3 deficiency. S. of essential athrombia. S. of thrombopathy.

Syndromography

There is a tendency to hemorrhage.

Paraclinical examinations

Blood. The adhesiveness, aggregation and release of platelet factor 3 is normalized in the presence of adenosine diphosphate (ADP).

Normality of the rest of the tests.

Syndromogenesis and etiology

It is a qualitative abnormality of platelets.

Of genetic etiology, autosomal recessive inheritance has been proposed.

Literature

Inceman, S.; S. Ucar and O. N. Ulutin: "Athrombia thrombocyto pathica". Thromb. Diath. Haemorrh., 4:234-243, 1960.

Weiss, H.J.: "Platelet aggregation, adhesion and adenosine diphospate release in thrombopathy (platelet factor 3 deficiency). A comparison with Glanzmann's thrombasthenia and Von Willebrand's disease." Amer. J. Med., 43:570-578, 1967.

BRUTON SYNDROME

Synonymy

S. of congenital sex-linked agammaglobulinemia.

Syndromography

Clinic

This syndrome appears only in men. Symptoms appear at 5 or 6 months of age and are characterized by be recurrent and accompanied by severe bacterial but not viral infections. The clinical picture is reminiscent of rheumatoid arthritis.

Paraclinical examinations

Blood. Almost complete absence of serum immunoglobulins. Cellular immunity is normal.

Medulogram. Absence of plasmacellen.

Syndromogenesis and etiology

There is an absence of plasma cells in the presence of a normal thymus. There are no tonsils, appendix, or Peyer's patches.

Pre B cells are present in the bone marrow but appear blocked in their ability to mature and express surface immunoglobulins or antibody production.

It is a genetic etiology syndrome of recessive inheritance linked to the X chromosome and the gene appears to be in Xq21.3-22.

Literature

Bruton, O.C.: “Agammaglobulinemia.” Pediatrics, 9:722-728, 1952.

Sell, S.: "Immunological diseases." Arch. Path., 86:95-107, 1968.

Malcolm, S. et al.: "Close linkage of sentom DNA fragments from Xq21.3-22 to X linked agammaglobulinaemia (XLA)." Human. Genet., 77:172-174, 1987.

CHEDIAK-HIGASHI SYNDROME

Synonymy

S. de Chediak-Steinbrinck-Higashi. S. of abnormal leukocyte inclusions accompanied by constitutional stigmata.

Syndromography

Clinic

This syndrome is characterized by photophobia and nystagmus on exposure to light. Weakness. Fever. Cerebellar tremor.

Dysmetria. Paralysis.

There is decreased pigmentation of the skin, hair and eyes. Generalized hyperhidrosis with a marked decrease in tear secretion, which can lead to corneal opacity.

Physical examination. Ruddy and painful nodules. Pustules and ulcerations of the skin. Moderate hepatomegaly.

Occasional splenomegaly and lymphadenopathy. Albinism in the fundus of the eye.

Paraclinical examinations

Blood. Anomalies are observed in the nuclear structures and granulations of all types of leukocytes: giants, cytoplasmic inclusions. Döhce bodies. Anemia.

Thrombocytopenia.

Medulogram. Large eosinophilic inclusions in myeloblasts and promyelocytes.

Syndromogenesis and etiology

The giant pleomorphic granules in leukocytes contain peroxidase and α-glucuronidase, but not alkaline phosphatase. They are considered giant lysosomes. It is transmitted by autosomal recessive inheritance.

The etiology is genetic.

Literature

Chediak, M.: "Nouvelle anomalie leucocytaire de caractere constitutionel et familial. " Rev. Hemat.. 7:362-367, 1952.

Weary, P.E., and A.S Bender: "Chediak-Higashi syndrome with severe cutaneous involvement, occurrence in two brothers 14 and 15 years of age". Arch. Int. Med., 119:381-386, 1967.

CHRISTMAS SYNDROME

Synonymy

S. of factor IX deficiency. S. deficiency of the plasma thromboplastin component (PTC). Hemophilia type B.

Syndromography

Clinic

This syndrome is virtually limited to men.

The name Christmas disease derives from the name of the 5-year-old boy who was the first case in which the syndrome was individualized. Hemorrhagic manifestations occur through natural orifices (hematemesis, melena, gingivorrhagia, hematuria, etc.). Post-trauma or spontaneous hematomas.

Post-tooth extraction hemorrhage.

Paraclinical examinations

Blood. Clotting time: prolonged or normal. Thromboplastin generation time: abnormal. PTC: altered.

Residual prothrombin: normal or altered. Determination of factor IX levels: decreased.

Normal bleeding time and prothrombin.

Syndromogenesis and etiology

There is a congenital deficiency of factor IX that is due to a recessive genetic disorder linked to the X chromosome.

This syndrome has been rarely observed in women.

Literature

Aggeler, P.M., et al.: "Plasma thromboplastin component (PTC) deficiency; a new disease resembling hemophilia." Proc. Soc. Exper. Biol. Med., 79:692-694, 1952.

Aggeler, P.M.; M.S. Hoag; R.0. Wallerstein, et al.: "The mild hemophilias: occult deficiencies of ANE, PTC, and PTA frequently responsible for unexpected surgical bleeding." Amer. J. Med., 30:84-94, 1961.

CONGENITAL INTRINSIC FACTOR DEFICIENCY SYNDROME

Syndromography

Clínica

It affects both sexes and sets in at an early age. There is paleness of the skin and mucosa, and there are no manifestations of malabsorption.

Paraclinical examinations

Blood. Megaloblastic anemia, demonstrated by blood count and medullogram.

Gastrochymogram. Normal.

Biopsy of the gastric mucosa. Absence of atrophy.

Schilling test. Positive.

Syndromogenesis and etiology

There is an absence of the hereditary intrinsic factor and a genetic etiology has been proposed for this defect.

Literature

Herbert, V.; R.R. Streiff and L.W. Sullivan: *Notes on vitamin B12 absorption: autoimmunity and pernicious anemia: relationship or intrinsic factor to blood group substance". Medicine, 43:679- -687, 1964.

FACTOR VII DEFICIENCY SYNDROME

Synonymy

S. of proconvertin deficiency. S. of prothrombin deficiency.

Syndromography

Clinic

It affects both sexes, but predominates in women. Its onset is variable from birth to adulthood. In some cases it is asymptomatic. There are spontaneous epistaxis, hematomas. Genitourinary and gastrointestinal hemorrhages.

Hemarthrosis. Hemorrhage associated with trauma or surgery.

Paraclinical examinations

Blood. Prolonged prothrombin time. It is corrected after adding serum. Prothrombin consumption. Normal clotting time. Generation of normal thromboplastin. Normal bleeding time.

Syndromogenesis and etiology

It is due to a congenital deficiency of factor VII. It presents autosomal recessive inheritance and heterozygotes may have minor manifestations.

The mutated factor VII gene has been mapped to 13q34.

Literature

Alexander, B.; R. Goldstein; G. Landwehr, et al.: "Congenital SPCA deficiency." J. Clin. Invest., 30:596-608, 1951.

FACTOR X DEFICIENCY SYNDROME

Synonymy

Stuart factor deficiency S. Stuart-Power factor deficiency S.

Syndromography

Clinic

It affects both sexes. Its onset varies from birth to adulthood. Other times it is asymptomatic. in natural orifices (gastrointestinal, urinary). There are epistaxis, hematomas, hemorrhagic manifestations in natural orifices (gastrointestinal, urinary). Posttrauma or surgery.

There are few manifestations in heterozygotes.

Paracliric exams

Blood. Prolonged prothrombin time. Altered prothrombin consumption. ST and PVEW (viper venom), abnormal. Altered thromboplastin generation time.

Decreased factor X quantification.

Syndromogenesis and etiology

There is congenital deficiency of factor

Literature

Gilgenkrantz, H., et al.: "Structural genes of coagulation factor VII and X located on 13q34." Ann. Genet., 29:32-35, 1986.
Hougle, C.; E.M. Barrow and J.B. Graham: "Stuart cloting defect." J. Clin. Invest., 36:485-497, 1957.

FACTOR XI DEFICIENCY SYNDROME

Synonymy

S. of Rosenthal factor deficiency. S. of PTA factor deficiency. S. of hemophilia C.

Syndromography

Clínic

It affects both sexes. It presents manifestations similar to those of hemophilia. Spontaneous hemorrhages, as well as purpura or hemarthrosis, are rare. Generally, manifestations secondary to trauma or post-surgery occur. The latter occur 2-3 days after the interventions. There are severe forms.

Paraclinical examinations

Blood. Normal bleeding time. Slightly prolonged or normal clotting time. Increase in partial thromboplastin time that can be corrected by normal plasma or non-fresh serum. Abnormal generation of thromboplastin.
Abnormal prothrombin consumption. Normal prothrombin time. Decreased factor XI quantification.

Syndromogenesis and etiology

There is a congenital deficiency of factor XI that is transmitted by autosomal recessive inheritance.

The gene (F1I) has been located at 4g35 and three types of mutations have been detected, however the genotype has the same genotype.

Literature

Asakai, R. et al.: "Factor XI (plasma thromboplastin antecedent) deficiency in Ashkenazi Jews in a bleeding disorder that can result from three types point mutations." Proc. Nat. Acad. Sci., 86:7667-7671, 1989.
Rosenthal, R.L.; 0.H. Dreskin and N. Rosenthal: "New hemophilia-like disease caused by deficiency of a third plasma thromboplastin factor." Proc. Soc. Exper. Biol. & Med., 82:171-174, 1953.

FACTOR XI DEFICIENCY SYNDROME

Synonymy

Hageman factor deficiency S.

Syndromography

This syndrome is generally asymptomatic, it is discovered in hospitals through routine examination prior to surgical treatments.

Paraclinical examinations

Blood. Normal bleeding time. Prolonged coagulation time. Long PTT. Abnormal P.G.T. Altered prothrombin consumption. Normal prothrombin time.

Syndromogenesis and etiology

It is a congenital deficiency of factor XII and is transmitted by autosomal recessive inheritance.

Literature

Ratnoff, O.D. and J.E. Colpy: Familial hemorrhagic trait associated with deficiency of clot-promoting fraction of plasma." J. Clin. Invest., 34:602-613. 1953.

FACTOR XII DEFICIENCY SYNDROME

Synonymy

S. of fibrin-stabilizing factor deficiency.

Syndromography

Clínic

Patients bleed shortly after the clot has occurred. Bleeding from the umbilical cord is observed in the newborn. It is proposed that the lack of this factor causes a delay in wound healing and predisposes to postoperative eventrations.

Rumpel-Leede test positive.

Paraclinical examinations

Blood. Noticeably prolonged bleeding time. Qualitative determination of factor XII must be performed (5 molar urea or 2% acetic acid technique). With this technique the deficiency can be demonstrated.

Syndromogenesis and etiology

This deficiency may be congenital or secondary to liver disease, intravascular coagulation or fibrinolysis.

In the first case, it is transmitted by autosomal inheritance. recessive.

Literature

Baggett, R.T.; J.W. Hampton and R.M. Bird: “Fibrinogen bleeding complicated by factor XIII defect." Arch. Int. Med., 121:539- -544. 1968.

Koller, E: Fibrinogen and fibrin turnover of clotting factors Thromb. Diath. Haemorrh., suppl. 13, pp. 109 and 411, 1963.
Robbins, K.C.:A study on the conversion of fibrinogen to fibrin. Amer. J. Physiol., 142:581-588, 1944.

GLUTATHIONE DEFICIENCY SYNDROME

Syndromography

Clinic

This syndrome affects both sexes. It usually begins in infancy or childhood. Chronic hemolysis may occur

acute hemolytic crisis spontaneous or induced by oxidant drugs.

There are neurological manifestations.

Paraclinical examinations

Blood. Decrease in glutathione reductase in erythrocytes.

Heinz bodies. There is hemolytic type anemia.

Syndromogenesis and etiology

The fundamental defect is the deficiency of glutathione-reductase in erythrocytes, leukocytes and platelets.

This syndrome is transmitted by autosomal dominant inheritance.

nante.

The etiology is genetic.

Literature

Waller, H.D.; W. Kaufmann:; W. Gerok, et al.: Biochemimische Befunde bei heterozygoten Glutathionreduktaseman gel-krägerinnen Pilvergiftung. Klin, Wchr., 42:613-618, 1964.

DEFIBRINATION SYNDROME

Synonymy

S. of acquired hypofibrinogenemia.

Syndromography

Clínic

Hemorrhagic manifestations occur associated with various pathologies: cyanotic congenital heart disease. liver disease, amyloidosis, purpura fulminans, Waterhouse-Friederichsen, giant cavernous hemangioma, extracorporeal circulation, septic abortion, stillbirth syndrome, amniotic embolism, placenta previa, leukemias. It is also associated with fibrinolysis, lung and prostate surgery.

Paraclinical examinations

Blood. Low fibrinogen. Prolonged prothrombin time. Decreased platelet count. Clotting time may be prolonged depending on the severity of the condition. When associated with fibrinolysis there are alterations in clot lysis. Euglobulin lysis. PTT altered. Prolonged prothrombin time. Increased degradation products.

Syndromogenesis and etiology

In defibrination syndrome there is consumption of factors I, II, V, VIII and platelets, and it is triggered by several disorders that produce excessive activation of coagulation.

This syndrome is usually associated and secondary to various pathologies:

A. Deficient clot formation due to fibrinogen deficiency with an increase in hematocrit:

1. Congenital heart disease.
2. Polycythemia true.

B. Alteration in fibrinogen synthesis:

1. Liver diseases.

C. Due to excess use:

1. Massive venous thrombosis.
2. Fulminating purple.
3. Incompatible blood.
4. Septic abortion.
5. Neoplasms.
6. Extracorporeal circulation.
7. Waterhouse-Friederichsen syndrome.
8. Placenta previa.
9. Dead fetus syndrome.

D. Primary and secondary fibrinolysis.

Literature

Fletcheh, A.P.; N. Alkjaersig and S. Sherry: "Pathogenesis of the coagulation defect developing during pathological plasma proteolytic (fibrinolytic) states. I. The significance of fibrinogen proteolysis and circulating fibrinogen breakdown products." J. Clin. Invest., 41:896-916, 1962.

Sharp, A.A.: "Pathological fibrinolysis." Brit. Med. Bull., 20:240-245, 1964.

DIGUGLIELMO SYNDROME

Synonymy

S. of erythroleukemia. S. of erythremia. S. of erythro-blastomatosis. S. of erythromyelosis.

Syndromography

Clinic

Acute form: asthenia, dyspnea on exertion, gingivorrhagia, fever. Pale skin and mucous membranes, petechiae and ecchymosis. There is hepatosplenomegaly with foci of myeloid metaplasia and lymphadenopathy.

Chronic form: weakness, refractory chronic anemia.

Splenomegaly generally found before the appearance of nucleated red blood cells in peripheral blood.

Paraclinical examinations

Acute form:

Blood. Nucleated erythrocytes (erythroblasts) are observed in different stages of maturation. Normochromic anemia with anisocytosis, poikilocytosis, Jowell-Jolly bodies, thrombopenia with hemorrhagic tendency and leukocytosis.

Medulogram. Erythroid elements predominate with dysplastic and anaplastic changes. Decrease in the number of megakaryocytes.

Chronic form:

Blood. Anemia, with anisocytosis, poikilocytosis, siderocytes.

Medulogram. Hyperplasia of the erythropoietic system with frequent megaloblastic changes. There is erythrophagocytosis.

Syndromogenesis and etiology

There is infiltration of erythroid elements in the spleen, lymph nodes, liver, heart and other organs.

It is considered a leukemic-type malignant condition of the erythropoietic system.

For both forms the etiology is unknown.

Literature

(di)Guglielmo, G.: A case of erythroleukemia." Folia Med., 3:319, 1917.

Magalini, S.L. and L. Alstrom: Clinical and hematologic aspects of acute erythromyelosis." J. Pediat., 52:501-530, 1958.

DOAN-WISEMAN SYNDROME

Synonymy

S. of primary splenic neutropenia.

Syndromography

Clínica

It is characterized by fever and pain in the left upper quadrant. Hemolytic Îcterus, anemia. Ulcerations of the mouth and pharynx similar to those observed in agranulocytosis.

Various degrees of splenomegaly. After splenectomy, all manifestations return.

Paraclinical examinations

Blood. Marked neutropenia and thrombopenia. Determination of anti-leukocyte antibodies. Hyperchromic macrocytic anemia, reticulocytosis. Unconjugated hyperbilirubinemia.

Medulogram. Functional hyperplasia of all systems.

Syndromogenesis and etiology

There is hyperfunction and trapping of granulocytes in the spleen. It is often associated with other processes of splenic hyperfunction of erythrocytes and platelets.

The etiology is unknown.

Literature

Wiseman, B.K. and C. A. Doan: Primary splenic neutropenia. A. newly recognized syndrome closely related to congenital hemolytic icterus and essential thrombocytopenic purpura." Ann. Int. Med., 16:1097-1117, 1942.

Doan C.A. and C.S. Wright: "Primary congenital and secondary acquired splenic panhematopenia." Blood, 1:10-26, 1946.

D0AN-WRIGHT SYNDROME

Synonymy

S. of splenic panhematopenia. S. of primary splenic pancytopenia.

Syndromography

Clinic

It affects both sexes and all ages. Acute or gradual installation. There are secondary manifestations of anemia, thrombocytopenia and neutropenia. Splenomegaly.

Paraclinical examinations

Blood. Anemia, variable reticulocytosis, neutropenia, thrombocytopenia. Hyperbilirubinemia.

Coombs test. Negative.

Medulogram. Hyperplasia of all series.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Doan, C.A. and C.s. Wright: “Primary congenital and secondary acquired splenic panhematopenia." Blood, 1:10-26, 1946.

DONATH-LANDSTEINER SYNDROME

Syndromography

Clinic

It affects both sexes and is established in all ages, the syphilitic type is established in the group of young people.

Paroxysmal attacks of hemoglobinuria occur. In some patients there are frequent episodes induced by minor exposure to cold and in other rare cases induced only by exposure to very low temperatures. In a period of minutes to hours after exposure to cold and also limited to a single part of the body, episodes of pain in the back and legs, abdominal cramps, headache, chills and hyperpyrexia occur and hours later hemoglobinuria, temporarily splenomegaly and jaundice appear accompanying each crisis. Other times, episodes of simple hemoglobinurias without systemic manifestations may occur. There are vasomotor phenomena. Acroparesthesia, urticaria. Acrocyanosis.

Raynaud's phenomenon with or without gangrene in some cases (S. of pseudo-Raynaud).

Paraclinical examinations

Blood. Anemia after attacks and alterations in laboratory tests in patients with hemolytic crisis.

Leukopenia followed by neutrophilic leukocytosis.

Donath-Landsteiner test. Positive.

Rosembach test (immersion of the limb in ice water). Positive.

Serology to rule out syphilis.

Urine. There is hemoglobin and methemoglobin.

Syndromogenesis and etiology

This syndrome can be caused by congenital (chronic type) and idiopathic (acute and chronic transient form) syphilis or associated with the presence of cold agglutinins.

The idiopathic form is probably due to an enzymopathy and is characterized by the reticulocytes containing less than 50% acetylcholinesterase.

Literature

Elliotson, J.: Diseases of the heart united with ague'". Lancet 1:500-501, 1832.

Dacie, J.V.: The Haemolytic Anaemias. New York, Grune & Stratton, 1963.

FAMILIAR BENIGN ERYTHROCYTHEMIA SYNDROME

Synonymy

S. of familial erythrocytosis. Benign familial polycythemic S.

Syndromography

Clinic

It occurs in both sexes. Generally this syndrome is discovered in childhood and is asymptomatic.

Splenomegaly of moderate size is present in 50% of cases. It may be associated with congenital disorders.

Paraclinical examinations

Blood. Erythrocytosis, increased volume. Normal white blood cell count and platelets.

Syndromogenesis and etiology

There is some evidence that it is an autosomal dominant hereditary syndrome that may be due to a mutation in the regulation of 2-3-diphosphoglycerate.

Literature

Bernstein, J.: "Three cases of polycythemia rubra." West London Med. J., 19: 207-208, 1914.

Davey, M.G.; J.R. Lawrence; H. Lander, et al.: "Familial erythrocytosis, a report of two cases, and a review." Haemat Act. (Basel), 39:65-74, 1968.

Stamatoyannopoulos, G.: "Familial erythrocytosis Birth Defects Orig." Art. Ser., 8(3):39-45, 1972.

STRESS-DAMESHEK SYNDROME

Synonymy

S. of familial hypoplastic anemia without congenital defects.

Syndromography

Clinic

Appears at an early age. There is asthenia, epistaxis, and easy bleeding. Little development. Pallor. Ecchymosis, petechiae.

There is no hepatosplenomegaly or lymphadenopathy.

Paraclinical examinations

Medulogram. There is quantitative hypoplasia with normal qualitative development of its cells.

Blood. Progressive hyperchromic macrocytic anemia, leukopenia, thrombocytopenia, reticulocytosis.

Medulogram. Hypoplastic or aplastic marrow with fatty infiltration.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Estren, S. and W. Dameshek: "Familial hypoplastic anemia of childhood." Amer. J. Dis. Child., 73:671-687, 1947.

EVANS SYNDROME

Synonymy

S. of autoimmune thrombocytopenic purpura with idiopathic hemolytic anemia.

Syndromography

Clinic

It is the association of an idiopathic hemolytic anemia with Werlhof syndrome.

It may or may not have purpuric manifestations and may occur concomitantly or in succession, in the absence of an etiological factor.

Paraclinical examinations

Blood. Hemogram. Reduced hemoglobin, increased reticulocytes, thrombocytopenia.

Bilirubin. Increased at the expense of indirectness.

Serum iron. High.

Coombs test. Positive.

Syndromogenesis and etiology

It is considered to be produced by a hemolytic component caused by an autoimmune process, antibodies against the thrombocyte and erythrocyte system, with marked blood destruction.

Literature

Evans, R.; K. Takahashi; R.T. Duane, et al.: Primary thrombocytopenic purpura and acquired hemolytic anemia. Arch. Int. Med., 87:48-65, 1951.

Silverstein, M.N.; THE. Aaro and R.D. Kempers: "Evan's syndrome and pregnancy." Amer. J. Med. Sci., 252:106-111, 1966.

EWING SYNDROME

Synonymy

S. of endothelial myeloma. Undifferentiated cell sarcoma of bone.

Syndromography

Clinic

80% of cases occur under the age of 30 and predominate in men. It can begin in childhood or adolescence.

There is intermittent pain in any bone, more common in long bones or the pelvis, which intensifies as the disease progresses. Slight fever.

Physical examination. On palpation there is edema and increased skin temperature. Pain with pressure on the bones. Bone tumors are palpated and are visible.

Paraclinical examinations

Bone scan.

Marrow biopsy.

Blood. Leukocytosis, secondary anemia and elevated erythrocyte sedimentation rate.

Radiology. Subperiosteal tumors. Spontaneous fractures.

Syndromogenesis and etiology

There is proliferation of endothelial cells probably derived from marrow reticulum cells.

The etiology is unknown.

Literature

Boyer, C.W.; T.J. Bricker and R.H. Perry: "Ewing's sarcoma." Cancer, 20:1602-1606, 1967.

Ewing, J.: "Review and classification of bone sarcomas." Arch. Surg., 4:485-533, 1922.

BLACKWATER FEVER SYNDROME

Synonymy

S. of hemoglobinuric fever.

Syndromography

Clinic

The most common symptom is vomiting, in severe cases there is shock.

Dark red or black urine suddenly appears after the administration of quinine in patients with Falciparum malaria. There is jaundice, thready pulse, tachycardia, hypotension.

Paraclinical examinations

Blood. Severe anemia, which can reach 1,000,000 red blood cells after attacks.

Urine. Hemoglobinuria. Hyaline and granular cylinders and amorphous sediment with different pigments.

Syndromogenesis and etiology

The administration of quinine is a precipitating factor in the clinical manifestations of the disease.

Literature

Beeson, P.B. and W. McDermott: Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia, Saunders, 1967.

Cummings. J.C.: "Miasmatic haematuria." N. Orl. M. News & Hosp. Gaz., 6:811, 1859-1860.

GAISBÖCK SYNDROME

Synonymy

Hypertonic polycythemia S. S. of stress polycythemia. S. of benign polycythemia. S. of relative polycythemia. S. of pseudopolycythemia.

Syndromography

Clínic

This syndrome predominates in men and smokers. It settles in around 45 years. It is accompanied by nervousness.

Physical examination. Patients have a plump, pink appearance. They are moderately overweight. Slight conjunctival congestion. Some have persistent hypertension. There is hypertrophy of the left heart, the arteries and veins of the head and hands are very marked. There is no splenomegaly. They frequently present vertigo and headaches.

Paraclinical examinations

Blood. Determination of volume. There is a relative polycythemia (decreased plasma) or a moderate and absolute polycythemia (7-10 million erythrocytes). High hematocrit.

Leukocytosis, normal platelets.

Syndromogenesis and etiology

There is a constitutional predisposition, or one associated with emotional tensions. Various hypotheses are proposed:

1. Symptomatic polyglobulia in the course of hypertension.
2. Disorders of the vegetative system.

It must be differentiated from Vaquez-Osler syndrome.

Literature

Gaisböck, E: "Die Bedeutung der Blutdruckmessung fur die arz tliche Praxis." Deutsch. Arch. Klin. Med., 83:363-409, 1905.

Russell, R.P. and C.L. Conley: "Benign polycythemia: Gaisböck's syndrome." Arch. Int. Med., 114:734-740, 1964.

GARDENER-DIAMOND SYNDROME

Synonymy

S. of autoerythrocytic sensitization. S. purpura due to sensitization to the erythrocyte membrane.

Syndromography

Clínic

All patients are women. Several times the symptoms begin after an operation related to the reproductive organs.

There is a history of episodes of bleeding of different nature, prior to the onset of the manifestations. Patients present local burning that precedes the development of purpuric lesions by 18-24 hours, which remain painless for 5-7 days.

It generally affects the limbs, face, rarely the back. It is accompanied by psychological manifestations.

Physical examination. Purpuric lesions are observed in areas not exposed to trauma.

Paraclinical examinations

Blood. Normal platelet count and rest of the coagulogram. Injection of erythrocytes or erythrocyte stroma gives a positive response.

Syndromogenesis and etiology

There is autosensitization to erythrocytes or their stroma, possibly due to phosphatidylserine. Emotional factors can influence the clinical picture.

Literature

Gardener, F.H. and L.K. Diamond: “Autoerythrocyte sensitization: A form of purpura producing painful bruising following autosensitization to red cells in certain women." Blood, 10:675-690, 1955.

Groch, G.S: S.C. Finch: W., Rogoway and D.S. Fisher: "Studies in the pathogenesis of autoerythrocyte sensitization syndrome." Blood, 28: 19-33, 1966.

ACUTE CEREBRAL GAUCHER SYNDROME

Synonymy

Acute neuropathic Gaucher syndrome. S. de Gaucher acute infantile. S. de Gaucher malignant. S. de Gaucher I.

Syndromography

Clinic

It affects both sexes although it is a little more common in men.

It appears between 6 months and 1 year after birth, less frequently in the newborn, and is accompanied by anorexia.

Physical examination. Progressive hepatosplenomegaly, strabismus, head hyperextension, hypertonicity, neck rigidity, dysphagia, apathy or catatonia, deep hyperreflexia, laryngeal spasm, mental retardation. Peripheral lymphadenopathy. Occasional seizures and lockjaw.

Paraclinical examinations

Blood. Increase in glycolipids, decrease in lipids and lipoproteins; anemia, leukopenia, thrombocytopenia (secondary to hypersplenism and bone marrow infiltration); elevated acid phosphatase.

Medulogram. Gaucher cells.

bone radiology. Typical destructive lesions in long bones.

Node biopsy. It is diagnostic and shows Gaucher cells.

Syndromogenesis and etiology

Abnormal accumulation of glucocerebrosides in reticuloendothelial cells due to enzymatic defect in the degradation of cerebrosides.

It is transmitted by autosomal recessive inheritance. It is a mutation of the same gene involved in Gaucher type I.

Literature

Frederickson, D.S.: Cerebroside Lipidosis: Gaucher's Disease. In Stanbury, J. B., J.B. Wyngaarden, and D.S. Frederickson: The Metabolic Basis of Inherited Disease. ed. 3, New York, McGraw-Hill, 1966.

Gaucher, P.: De lépithelioma primitif de la rate. These (Paris), 1882.

ADULT CHRONIC GAUCHER SYNDROME

Synonymy

S. de Gaucher I non-neuropathic.

Syndromography

Clínic

It affects both sexes and is much more common in Jews. It occurs at any age and can occur without symptoms.
There may be bone pain, pathological fractures, hypersplenism.

Physical examination. Splenomegaly, skin pigmentation, pingueculae (first on the nasal side and then temporal side of both eyes).

Paraclinical examinations

See Acute Gaucher.

Radiology. The most affected bones are the long bones, the vertebrae, the ribs and the pelvis; The cortex of the lower end of the femur is enlarged, giving the appearance of an Erlenmeyer flask.

Syndromogenesis and etiology

It is a lysosomal defect for the degradation of glucocerebroside and, although the deficiency exists in all body cells, it only affects macrophages, as a consequence there is an enlargement of the spleen and liver and is often due to painful lesions.

The enzyme deficiency is at the level of beta-glucosidase glucocerebrosidase, which normally hydrolyzes glucocerebrosides into ceramide and glucose. The gene that codes for this enzyme is located on chromosome 1 and has 11 exons.

Mutations that have been discovered include point mutations in exons and splice sites, nucleotide insertions, gene-pseudogene crossover, and gene conversion.

Based on its functional effect and association in the Gaucher phenotype, it is possible to classify this mutation into two groups: mutations associated with the severe neuropathic form and those not associated.

The basic knowledge of this genetic defect of autosomal recessive inheritance has made it possible to move from symptomatic treatment (splenectomy, separation of fractures, infections, etc.) to non-neuropathic bone marrow transplantation and enzyme replacement, which has excellent results but is very expensive. Annually the cost per patient, for enzyme alone, is 3,822,000 USD.

Literature

Butler, E.: “Gaucher Disease: New molecular approaches to diagnosis and treatment.” Science, 256: 794-799, 1992.

Frederickson, D.S.: Cerebroside Lipidosis: Gaucher's Disease. In Stanbury, J.B.J. B. Wyngaarden, and D.S. Fredrickson: The Metabolic Basis of Inherited Disease. ed. 3, New York, McGraw-Hill., 1966.

Gaucher, P: De lépithelioma primitf de la rate. These (Paris), 1882.

GAUCHER SYNDROME TYPE I

Syndromography

Clinic

It is manifested by: bone and joint pain, fever, purpura, epistaxis, hemorrhagic infarctions, hematomas. Conjunctival pigmentation. Hepatosplenomegaly.

Paraclinical examinations

Blood. Anemia, thrombocytopenia, leukopenia.

Spleen and bone marrow biopsy. Abundant cells of Gaucher.

Syndromogenesis and etiology

Of genetic etiology as in type II, it is a mutation of the same gene as Gaucher type I.

GLANZMANN SYNDROME

Synonymy

S. of Glanzmann's thromboasthenia. S. of hereditary thromboasthenia.

Syndromography

Clinic

It affects both sexes and is present from birth.

It can be asymptomatic until a trauma or a surgical procedure makes the clinical picture evident, which is characterized by recurrent petechiae, epistaxis, menorrhagia or metrorrhagia and other hemorrhages of different degrees of severity. The loop test can be positive or negative. This condition can also occur spontaneously. In general there is no splenomegaly.

Paraclinical examinations

Blood. Prolonged bleeding time. Normal or low platelet count, these may have a normal morphology, or be small or elongated and of various shapes, sometimes giant and with the absence of granulations and vacuoles. Platelets do not aggregate in the presence of adenosine diphosphate or pyruvatokinase. In a second variety, ATP is low and in a third variety, ATPase is deficient. In a fourth variety there is an absence of a fibrinogen-like protein.

Coagulation tests. In general they are normal but clot retraction is slow or absent.

Thromboelastogram. Diminished.

Syndromogenesis and etiology

In this syndrome there is a qualitative defect in the platelets, which brings with it a false mechanism to produce viscous metamorphosis and clot retraction.

There is a specific enzymatic defect in the glycolytic system of platelets which is the lack of glyceraldehyde-3-phosphate dehydrogenase.

Four different genotypes have been identified that lead to the phenotype of this syndrome.

Different varieties of the syndrome have been considered that can transmit the trait by autosomal recessive or dominant inheritance.

Literature

Glanzmann, W.E.: "Hereditare hamorrhägische Thrombasthenie." Janrb. F. Kinderh., 88:1-42, 113-141, 1918.

Glanzmann, E., and P. Riniker: "Essentielle Lymphocytophise. "Ein neues Krankheitsbild aus der Säuglingspathologie". Ann. Pediat. (Basel), 175:1-32, 1950.

Ratnoff, O.D.: Hereditary Disorders of Hemostasis. In, Stanbury, J.B., J.B. Wyngaarden, and D.S. Fredrickson.: The Metabolic Basis of Inherited Diseases. ed. 2, New York, McGraw-Hill, 1967.

HEGGLIN SYNDROME

Synonymy

S. of May. May-Hegglin anomaly S.

Syndromography

Clinic

Generally, there is no deterioration in health, only the existence of minimal hemorrhagic manifestations.

Physical examination. There are no signs, or paleness and hemorrhagic manifestations

Paraclinical examinations

Blood. Moderate leukopenia, increased percentage of polymorphonuclear cells, which show inclusions cytoplasmic or shape disorders. Thrombocytopenia and giant platelets. Prolonged clot retraction. Positive loop test. Doehle corpuscles are characteristic in mature, basophilic nuclear polymorphs and in monocytes.

Syndromogenesis and etiology

Congenital disease, of genetic etiology and autosomal dominant inheritance, with variable expression.

The etiology is unknown.

Literature

Davis, J.W. and S.J. Wilson: "Platelet survival in the May-Hegglin anomaly." Brit. J. Haemat., 12:61-65, 1966.

Hegglin, R.: "Gleichzeitige konstitutionelle Veranderungen an Neutrophilen und Thrombozyten." Helvet. Med. Acta, 12:439-440, 1945.

CLASSIC HEMOPHILIA SYNDROME

Synonymy

S. of hemophilia A. S. of factor VII deficiency. S. of antihemophilic globulin deficiency.

Syndromography

Clinic

It appears in men. It is sometimes installed at birth. There is bleeding from the umbilical cord or after circumcision. Hemorrhages occur in the mucous membranes, digestive system, lungs, intracranial, and genitourinary areas. Hemarthrosis. Haemo-Severages secondary to dental extractions. Bruises from trauma.

Paraclinical examinations

Coagulation time. Normal or prolonged.

Partial thromboplastin time (PTT). Altered.

Residual prothrombin. Altered

Prothrombin time. Normal.

Factor quantification. Factor VIII deficiency.

Lack of antihemophilic factor in serum.

Bleeding time. Normal.

Syndromogenesis and etiology

There is a congenital deficiency of factor VIII.

It is a recessive disease linked to the X chromosome.

Literature

Otto, J.C.: "An account of an hemorrhagic disposition existing in certain families." M. Repository, 6:1, 1803.

Shulman, N.R.; D.H. Cowan; E.P. Libre, et al.: "The physiologic basis for therapy of classic hemophilia (factor VIII deficiency) and related disorders." Ann. Int. Med., 67:856-882, 1967.

HEMOGLOBIN C SYNDROME IN HOMOZYGOTHES (CC)

Syndromography

Clinic

There is recurrent, moderate pain in the bones and joints. Recurrent abdominal pain of varying intensity, seizures, hemorrhagic manifestations.

Physical examination. Jaundice. Splenomegaly. There are no skeletal abnormalities.

Paraclinical examinations

Blood. Moderate normocytic or microcytic anemia, microspherocytes, target cells, reticulocytosis, moderate hyperbilirubinemia, decreased osmoic fragility. frequent count.

Hemoglobin electrophoresis. Type C.

Medulogram. Hyperplasia.

Syndromogenesis and etiology

Hemoglobin C is the result of a genetic abnormality due to a mutation of the beta-hemoglobin locus that determines the exchange of the glutamic amino acid for lysine. The heterozygous patient is asymptomatic, but the CC homozygous may present the syndromography described here. The beta-hemoglobin locus is located on chromosome 1lpl1-p15.

Inheritance is autosomal recessive.

HEMOGLOBIN C SYNDROME (TRAIT)

Syndromography

Clínica

Generally asymptomatic, occasionally bone and dental lesions have been seen. Unilateral renal hematuria.

Paraclinical examinations

Blood. There is no anemia. Presence of target cells and microspherocytes. There is a reduction in osmotic fragility. Presence of hemoglobin C in different percentages.

Syndromogenesis and etiology

In these patients there is a hemoglobin defect that is transmitted by autosomal recessive inheritance.

HEMOGLOBIN C THALASSEMIA SYNDROME

Synonymy

S. de Suelzer-Kaplan.

Syndromography

Clinic

Moderate or severe bone pain. Splenomegaly is strange.

Paraclinical examinations

Blood. There is anemia, target cells, schistocytosis, microspherocytosis. Decreased osmoic fragility.

Medulogram. cellular hyperplasia.

Syndromogenesis and etiology

It is a genetic defect resulting from a genotype composed of a C mutation (see hemoglobin C syndrome in homozygotes (CC)) and a thalassemic mutation (see thalassemia).

Literature

Itano, H.A.: “A third abnomal hemoglobin associated with hereditary hemolytic anemia." Proc. Nat. Acad. Sc., 37:775-784, 1951.

HEMOGLOBIN SYNDROME IN HOMOZYGOTS

Syndromography

Clínica

There is pallor, occasionally polycythemia, moderate splenomegaly or absence thereof.

Paraclinical examinations

Blood. Moderate normochromic microcytic anemia, target cells, decreased osmotic fragility and moderate hyperbilirubinemia.

Syndromogenesis and etiology

It is a mutation of the beta hemoglobin locus (glutamic 26 to lysine) that in a homozygous state can produce the symptoms described.

PAROXYSTIC COLD HEMOGLOBINURIA SYNDROME

Synonymy

S. of cryoglobulinemia.

Syndromography

Clinic

It generally begins between 30-80 years of age with a slight prevalence in women.

There is anemia, more pronounced in the winter. Raynaud's phenomenon is observed. Hemoglobinuria that is expressed by exposure to cold. There is pallor, jaundice, occasionally hepatos-plenomegaly. Purpuric lesions followed by hyperpigmentation lesions. Ankle ulcers. Acrocyanosis in the fingers that become stiff and slightly painful;
This can lead to gangrene especially with exposure to cold and improves in a warm environment.

Paraclinical examinations

Blood. There is anemia. The erythrocytes show fewer abnormalities when compared to those of the heat agglutination type. There is moderate reticulocytosis. Marked increase in autoagglutination. Normal or slightly increased osmotic fragility. Leukocytes and thrombocytes decreased, normal or increased. Variable hyperbilirubinemia.

Coombs test. Direct positive. Much increased cold agglutination. Decreased serum complement.

Urine. Increased urobilin. Hemoglobinuria associated with exposure to cold.

Stool. Increased fecal stercobilinogen.

Syndromogenesis and etiology

In many cases it is associated with infections such as primary atypical pneumonia (mycoplasma), viruses, collagen diseases or lymphomas. Mixed IgG-IgA- -IgM cryoglobulinemia has been reported and therefore it has been concluded that this essential mixed cryoglobulinemia has an autosomal dominant inheritance.

Literature

Hayem, G.: "Sur une varieté particuliére d'ictéré chronique; ictére infectieux chronique spléno-mégalique." Presse Méd. J.:121- -125, 1898.

Logothetis, J.; W.R. Kennedy:; A. Ellington, et al.: "Cryoglobulinemic neuropathy, incidence and clinical characteristics." Arch. Neurol., 19:389-397, 1968.
Nightingale, S.D. et al.: "Inheritance of mixed cryoglobulinemia." Am. J. Hum. Genet., 33:735-744, 1981.
Widal, E. and P. Abrami: "Types divers d'ictéres hémolytiques non congénitaux, avec anémie; la recherche de la résistance globulaire par le procédé des hématies déplasmatisées." Bull. Et Mem. Soc. Méd. D'Hop. de Paris, 24:1127-1 169. 1907.

MULTIPLE MYELOMA HEMORRHAGIC SYNDROME

Syndromography

Clínic

In patients with myeloma, hemorrhages from the mucous membranes, purpura, weakness, visual disturbances, retinal hemorrhages occur.

Paraclinical examinations

Medulogram. Infiltration of the marrow by plasma cells appears, which causes interference with the progenitor cells.

Radiology. Osteolytic areas can be seen.

Protein electrophoresis. The "M component of serum" appears.

Blood. Thrombocytopenia, functional thrombopathy. Prolonged prothrombin time. Low fibrinogen. Disorders in platelet aggregation.

Syndromogenesis and etiology

Bleeding disorders are secondary to thrombocytopenia caused by infiltration of plasma cells into the bone marrow. Platelet function disorders and vascular capillary infiltration.

The interaction of globulins with connective tissue is also considered.

The etiology is unknown.

Literature

Luscher, E. (Berne) and A. Labhart: "Blutgerinnungsstorung durch By-Globuline; zur-kenntnis der Gerinnungsstorungen durch korpereigene Antikoagulantiep. Schweiz." Med. Wschr. 79:598-604, 1949.
Stefani, M. and W. Dameshek: The Hemorrhagic Disorders, a Clinical and Therapeutic Approach. ed. 2, New York, Grune, 1962.
Vigliano, E.M. and HI. Horowitz: "Bleeding syndrome in a patient with Ig A myeloma: interaction of protein and connective tissue." Blood, 29:823-836. 1967.

HERMANSKY-PUDLAK SYNDROME

Synonymy

S. of thrombocytopathy, albinism and pigmented marrow cells.

Syndromography

Clinic

There is albinism and hemorrhagic manifestations like Glanzmann syndrome. The clinical picture is pseudohemophilia: hemorrhagic diathesis, prolonged bleeding time without thrombopenia or factor VII deficiency.

Syndromogenesis and etiology

Of genetic etiology. It is an autosomal recessive inherited condition whose basic defect is unknown although storage of a ceroid-like material in the tissues similar to the defect in neuronal ceroid lipofusinosis has been observed.

Literature

Hermanski, F. and P. Pudlak: Albinism associated with hemorrhagic diathesis and unusual pigmented reticular cells in bone marrow." Blood, 14:162-169, 1959.
Takashi, A. and T. Yokoyama: "Hermansky-Pudlak syndrome with special reference to lysosomal dysfunction: a case report and review of the literature." Virchows Arch. Path. Anat., 402:247-258, 1984.

HERRICK SYNDROME

Synonymy

Sickle cell anemia. S. of sickle cell anemia. S. of meniscocytosis. S. of Dresback.

Syndromography

Clinic

It is observed, particularly, in blacks, also in white families from Greece and Italy, especially Sicilians. It is more common in women. It is characterized by chronic anemia, moderate jaundice, rapid increase in fatigue, and severe joint pain. Abdominal cramps, especially in the epigastrium, right hypochondrium (which simulates an acute abdomen), fever during crises. There is spontaneous hematuria, epistaxis, neurological manifestations: headache, hemiplegia, aphasia, temporary or permanent blindness.

Physical examination. There is pale skin, moderate subicterus of the skin and mucous membranes, short trunk and elongated limbs, increased anteroposterior diameter of the thorax. Cardiomegaly, tachycardia, sinus arrhythmia, systolic thrill in the precordium, accentuation of the second sound are confirmed. Moderate lymphadenopathy and splenomegaly is rare, since these patients self-splenectomize. Chronic ulcerations occur in the lower limbs.

Paraclinical examinations

Blood. Anemia, with anisopoikilocytosis, some sickle cells, which are best demonstrated in the absence of oxygen, nucleated erythrocytes, increased reticulocytes. Leukocytosis during hemolytic crises.

Bosinophilia, leukocytosis with shift to the left. Increase in platelets. Increased osmotic fragility.
low erythrocyte sedimentation rate. Hyperbilirubinemia.

Huck test. Positive.

Medulogram. Hyperplasia of the erythropoietic system.
There may be megaloblastic changes.

Urine. Hematuria.

Radiology. Bony abnormalities in the skull (tower skull). Osteoporosis and osteosclerosis, aseptic necrosis of the heads of the femurs. Pathological fractures.

Syndromogenesis and etiology

It is a congenital hemoglobinopathy, of the autosomal type, where there is substitution of glutamic acid for the valine molecule in position 6 of the beta chain. Red blood cells take the form of bananas in the presence of an oxygen deficit.

There is commonly a hereditary variant of hemoglobin (Hb-S, Hb-C) demonstrable by electrophoresis.

Literature

Colombo, B.; E. Svarch and G. Martinez: Introduction to the study of hemoglobinopathies. Scientific-Technical Ed. City of Havana, 1981.

Herrick, J.B.: "Peculiar elongated and sickle-shaped red corpuscles in a case of severe anemia." Arch. Int. Med., 6:517-521, 1910.

HYPERVISCOSITY SYNDROME

Synonymy

S. rheological. Hyperglobulinemic purpura S.

Syndromography

Clínica

There are visual disorders, epistaxis, mucosal hemorrhages, vertigo, asthenia, anorexia, dyspnea, syncope and seizures.

Physical examination. In the fundus, sausage-like venous dilations and hemorrhages are seen. There is nystagmus, peripheral edema, and decreased pulse pressure.

Paraclinical examinations

Blood. Anemia. Normal leukocytes or increased lymphocytes. Normal platelets. Determination of viscosity.

Protein electrophoresis and other methods. To determine abnormal proteins in plasma.

Medullogram and lymph node biopsy. To rule out Waldeström disease, lymphomas.

Electroencephalogram. Shows specific changes.

Syndromogenesis and etiology

The syndrome is due to an increase in blood viscosity secondary to the presence of macroglobulins or other dysproteinemia.

Literature

Fahey, J.L.: "Serum protein disorders causing clinical symptoms in malignant neoplastic diseases." J. Chron. Dis., 16:703-712, 1963.

Fulton, J.E.; H.J. Hurley and R. Kennedy: *Purpura hyperglobulinemia". Arch. Derm., 97:446-449, 1968.

Solomon, A. and J.L. Fahey: "Plasmapheresis therapy in macroglo bulinemia." Ann. Int. Med., 58:789-800, 1963.

ACQUIRED HYPOPROTHROMBINEMIA SYNDROME

Syndromography

Clinic

It is characterized by hemorrhagic manifestations through natural orifices and ecchymosis that depend on the magnitude of the disorder.

Paraclinical examinations

Prothrombin time. Prolonged.

Coagulation time. It may be prolonged depending on the magnitude of the disorder. When there is a deficiency of vitamin K, there is a deficiency of prothrombin and plasma factors II, VII, and X.

In cases of liver disease there is a deficiency of prothrombin and factors II, V, VII, X and occasionally IX.

When Dicumarol-type anticoagulants are used, there is a deficiency of prothrombin, factors VII and sometimes discretely IX and X.

Syndromogenesis and etiology

It is due to a vitamin K deficiency, liver disease, and use of coumarin-type anticoagulants.

Bibliography

Josso, F; 0. Prou-Wartelle and J.P. Soulier: "Etude d'un Casd' Hypoprothrombinemie congenitale." Nouv. Rev. Fr. Hemat. 2:647-672, 1962.

Ratnoff, O.D.: "Hereditary Disorders of Hemostasis." In Stambury, J.B.; J.B. Wyngarden and D.S. Frederickson: The Metabolic Basis of Inherited Disease. ed. 2, New York, McGraw Hill, 1966.

CONGENITAL HYPOPROTHROMBINEMIA SYNDROME

Synonymy

S. of coagulation factor II deficiency. S. of prothrombin deficiency.

Syndromography

Clinic

It affects both sexes. It is generally installed from childhood. It presents symptoms very similar to those of classic hemophilia.

Paraclinical examinations

Prothrombin time. Prolonged 10% above normal control.

Coagulation time. Normal or prolonged.

Prothrombin time in a single time. Prolonged.

Quantification of factor I. Decreased.

Syndromogenesis and etiology

It is due to an autosomal recessive inherited prothrombin deficiency.

The etiology is unknown.

HODGKIN SYNDROME

Synonymy

Hodgkin's lymphogranuloma S. S. of Hodgkin's lymphosarcoma. S. of Pel-Ebstein. S. of Stemberg. S. of malignant granuloma. S. of Hodgkin's paragranuloma.

Syndromography

Clinic

It affects both sexes, men 67% and women 37%. There are three periods in terms of age: between 0-14, 15-34, 50 and more.

There is general malaise, anorexia, weight loss, nausea, fever that can be continuous, intermittent or Pel-Ebstein, accompanied by sweating, pruritus, cough, dysphagia, alcohol intolerance.

Physical examination. Paleness, lymphadenopathy of variable location: cervical (60-80%), axillary (6-20%), inguinal (6-12%), mediastinal (6-12%). etc., at first hard and painless.

Involvement of the lung, pleura. There is hepatospleno-megaly. Hemorrhages. The nervous system may be affected.

It can be associated with viral and fungal infections. Autoimmune hemolytic anemia, amyloidosis, collagen diseases.

Skin hyperpigmentation.

Paraclinical examinations

Node biopsy. It reveals the anatomopathological alterations that can take different aspects:

granulomas, proliferation of lymphocytes and/or histiocytes. Reed-Sternberg cells with infiltrates in the extracted tissue.

According to the histological characteristics, three categories have been considered: paragranuloma, granuloma and Hodgkin's sarcoma, which also imply different evolution and prognosis.

Blood. Anemia that can be hemolytic, with a positive Coombs test, although it is generally negative. Leukocytosis with neutrophilia, eosinophilia. Alkaline phosphatase in leukocytes elevated during the active phase. Accelerated erythrocyte sedimentation. Hypercalcemia. Hypophosphatemia. Protein electrophoresis: decreased albumin.

Radiology. There is lymphadenopathy. bone injuries.

Pyelography and splenoportography.

Syndromogenesis and etiology

It is debated whether it is a malignant neoplasm or a malignant inflammation.

The etiology is unknown

Literature

Hodgkin, T.: "On some morbid appearances of the absorbent glands and spleen." Doctor Chir. Trans., 17:68-114, 1832.

KAHLER-BOZZOLO SYNDROME

Synonymy

S. of multiple myeloma. S. of plasmacytoma multiple myeloma. S. de Huppert. S. of Mclntyre. S. de Rustitski.

Syndromography

Clinic

It occurs more frequently in men, generally after 50 years of age.

The main symptoms are: asthenia, anorexia, weight loss, persistent bone pain, more frequently in the back, less frequently in the chest and limbs. There are spontaneous fractures. Hepatomegaly may appear late. Splenomegaly is rare.

It is diagnosed by Kahler's triad: osteoporosis with spontaneous fractures, Bence-Jones proteinuria and cachexia.

Paraclinical examinations

Blood. Moderate anemia, normocytic, normochromic.

Normal leukocytes. Altered coagulation tests.

Monoclonal hypergammaglobulinemia, which affects the globulin fractions and characterizes the type of myeloma. Decrease in normal immunoglobulins. Accelerated erythrocyte sedimentation. Hypercalcemia. Normal match. Alkaline phosphatase normal or slight elevation. Hyperuricemia.

Medulogram. Multiple myelomatous cells, with notable individual variations.

Bence-Jones in urine. Positive (may be normal).

Radiology. Osteolysis zones. pathological fractures, osteoporosis.

Immunoelectrophoresis. Characterizes type of immunoglobulin.

Syndromogenesis and etiology

Abnormal proliferation of plasma cells with production of a paraprotein (myeloma protein).

Recently, some cases have been discovered that present aneuploid chromosomal anomalies.

The etiology is unknown.

Literature

Bozzolo, C.: "Sulla malattia di Kahler" Arch. Internag. say Med. e Chir., Napoli, 409, 1897.

Kahler, O.: “Zur Symtomatologie des multiples Myeloms; Beobachtung von Albumosurie". Prag. Med. Wschr., 14:33-45, 1889.

KASABACH-MERRITT SYNDROME

Synonymy

Capillary angioma syndrome with thrombocytopenic purpuric syndrome. S. of thrombopenia-hemangioma.

Syndromography

Clinic

It appears in the first months of life in the form of purpura and hemorrhages. There are giant hemangiomas, pallor, petechiae and ecchymoses.

Paraclinical examinations

Blood. Thrombocytopenia. Accumulation of platelets labeled with Cr 51 in the tumor. Anemia, increased fibrinolytic activity.

Medulogram. Disorders in the maturation of megakaryocytes.

Syndromogenesis and etiology

Angiomas cause sequestration of platelet thrombi with release of plasmin.

The mechanism of hematological changes is not known.

There is no evidence of a basic genetic defect.

Literature

Kasabach, H.H. and K.K. Merritt: "Capillary hemamgioma with extensive purpura: Report of a case." Amer. J. Dis. Child., 59:1063-1070, 1940.

Quick, A.J.: Hemorrhagic Diseases and Thrombosis. Philadelphia, Lea & Febiger, 1966.

CHRONIC HYPOPLASTIC LEUKOPENIA SYNDROME

Synonymy

S. de Spaet-Dameshek. S. of primary hypoplastic neutropenia.

Syndromography

Clínica

This syndrome usually appears between 15 and 65 years of age. There are repeated and prolonged infections of the skin, mouth,

ears, sinuses and lungs. Moderate splenomegaly may be found.

Paraclinical examinations

Blood. Severe neutropenia, occasionally moderate anemia and thrombocytopenia.

Medulogram. Hypoplasia of the granulopoietic system.

Syndromogenesis and etiology

The bone marrow presents selective neutrophilic hypoplasia.

The etiology is unknown.

Literature

Hattersley, P.G.: "Chronic neutropenia. Report of case not cured by splenectomy." Blood, 2:227-232, 1947.

Wintrobe, M.M.: Clinical Hematology, Philadelphia, Lea & Febiger, 1967.

AFRICAN LYMPHOMA SYNDROME

Synonymy

Burkitt lymphoma S. S. of non-leukemic lymphoma.

Syndromography

Clinic

African lymphoma has been detected in patients from the central region of equatorial Africa. It has been observed from childhood to adolescence.

Physical examination. A painless, rapidly growing tumor mass is observed in the jaw or salivary glands, cervical lymph nodes, or abdomen. Does not invade the skin.

Paraclinical examinations

Biopsy of the tumor. Proliferation of uniform round cells, intertwined with infiltration of reactive reticular cells that give the appearance of a starry sky.

Blood. There is no evidence of leukemia. Antibodies against the Epstein-Barr virus are detected in a high percentage of cases.

Syndromogenesis and etiology

A viral etiology has been considered from a hypothetical point of view. The pathogenetic mechanism is not clear. There is a translocation break at 8q24. The interaction of loci or genes is discussed; The MYC oncogene on chromosome 8 is under the control of the immunoglobulin promoter and overproduction of MYC occurs, which immortalizes the lymphocyte and a second mutational event of another RAS type oncogene triggers the disease.

The etiology is unknown.

Literature

Burkitt, D.: "A sarcoma involving the jaws in African children." Brit. J. Surg., 46:218-223, 1958.

Sacks, R.L.: "Burkitt's tumor (African lymphoma syndrome) in California". Oral Surg., 22:621-627, 1966.

HEREDITARY INTESTINAL MALABSORPTION SYNDROME OF VITAMIN B12

Synonymy

Hereditary megaloblastic anemia with proteinuria.

Syndromography

Clinic

It affects both sexes and sets in in the second year of life.

There is fatigue and weakness, pale skin and mucous membranes.

Paraclinical examinations

Blood. Megaloblastic anemia. Vitamin B12 deficiency.

Medulogram. Typical megaloblastic changes.

Gastrochymogram. Normoacidity and normality of the intrinsic tor.

Urine. Proteinuria and in some cases aminoaciduria.

Syndromogenesis and etiology

It is a methylcobalamin deficiency of genetic etiology that presents heterogeneity in affected patients. As it is presumably a congenital error in cobalamin metabolism, an autosomal recessive inheritance is proposed.

Literature

Grasbek, R.; R. Gordin; I. Kantero et al.: "Selectmalabsorption and proteinuria in young people: a syndrome." Acta Med. Scand., 167:289-296, 1960.

Grasbeck, R. and G. Kvist: "Kongenitale spezifische Vitamin-B12 Malabsorption mit Proteinurie." Munchen. Med. Wschr. 109:1936-1944, 1967.

Watkins, D. and D.s. Rosenblat: "Genetics heterogeneity among patients with methylcobalamin deficiency." J. Clin. Invest., 81:1690-1694, 1988.

MARCHIAFAVA-MICHELI SYNDROME

Synonymy

S. of paroxysmal nocturnal hemoglobinuria. Strübing-Marchiafava paroxysmal nocturnal hemoglobinuria.

Syndromography

Clinic

It occurs between the third and fourth decades. It affects both sexes. It is frequently asymptomatic or is manifested by attacks of lumbar and substernal pain, malaise and fever.

Physical examination. Paleness, yellowing of the skin and mucous membranes, sometimes tanning.

There are functional murmurs, splenomegaly, and occasionally hepatomegaly.

Renal hemosiderosis and hemoglobinuric nephrosis appear as late complications.

Paraclinical examinations

Blood. Anemia, reticulocytosis, hyperbilirubinemia, free hemoglobin in plasma. Leukopenia, alkaline phosphatase

low neutrophils, moderate thrombocytopenia. Low serum iron.

Medulogram. Erythrocyte hyperplasia.

Urine. Urobilinuria, hemosiderinuria and free hemoglobin in or1na.

Syndromogenesis and etiology

The syndrome is caused by an intracorpuscular erythrocyte defect of unknown etiology. Different infections can trigger crises.

It is a rare acquired disorder, resulting from an unusual susceptibility of the erythrocyte to the lytic action of complement, probably due to defects in specific proteins of the erythrocyte membrane (glycosyl-phosphatidylinositol) due to mutations of that gene, for which an autosomal recessive inheritance is proposed.

Literature

Low, M.G and A.R. Saltiel: Structural and functional roles of glycosylphosphatidylinositol in membranes. Science, 239:268-275, 1988.

Marchiafava, E. and A. Nazari: "Nuovo contributo allo degli itteri cronici emolitici", Policlinico (Sez. Prat.), 18:241-254, 1911.

Micheli, E: "A case of emolitic anemia with perpetual emosideriuria." Accad. di Med. Torino, 7:148, 1928.

Mengel, C.E.; H.E. Kann and W.D. Meriwether: “Studies of paroxysmal nocturnal hemoglobinuria erythrocytes: increased synthesis lipid peroxide formation by hydrogen peroxide." J. Clin. Invest., 46:1715-1723, 1967.

HEREDITARY METAHEMOGLOBINEMIA SYNDROME

Synonymy

St. Hörlein-Weber

Syndromography

Clínic

It affects both sexes from childhood. Sometimes discovered at a later age. Cyanosis is described (no clubbing or murmurs). Exercise can cause dyspnea, palpitations, and headache. The conjunctival and retinal veins may be congested. In some families it can be associated with mental deficiencies in other relatives.

Paraclinical examinations

Hemoglobin electrophoresis and spectrochemical determinations.

Determination of diphosphopyridine nucleotide.

Syndromogenesis and etiology

It is a defect of genetic etiology due to a mutation in the alpha or beta chains of hemoglobin that changes important amino acids in the chemical formation in which the heme group is located. Alterations in this site make it difficult for the erythrocyte to transport oxygen. There are different variants due to substitutions at this level of the alpha and beta chains.

Literature

Colombo, B.: E. Svarch and G. Martínez: Introduction to the study of hemoglobinopathies. Technical Scientific Editorial, City of Havana, pp. 216-221, 1981.

Gerald, P.S. and E.M. Scott: The Hereditary Methemoglobinemias. In Stanbury, J.B., J.B. Wyngaarden and D.S. Frederickson: The Metabolic Disease. ed. 2, New York, McGraw-Hill, 1966.

Himans van den Bergh, A. A. and A. Grutterink: "Enterogene cyanose." Berl. Klin. Wschr., 43:7-10, 1906.

MYELOFIBROSIS SYNDROME

Synonymy

S. de Harison-Vaughan. S. of Vaughan. S. of agnogenic myeloid metaplasia. S. non-leukemic chronic myelosis.

Syndromography

Clínic

It affects both sexes. It usually sets in insidiously after the fifth decade. Rare in children. It is characterized by weakness, fatigue, weight loss, anorexia. Moderate hemorrhagic manifestations. Discomfort in the left hypochondrium.

Physical examination. Pallor, splenomegaly. Frequent hepatomegaly. Rarely moderate lymphadenopathy.

Paraclinical examinations

Blood. Normocytic anemia, of variable degree. Polycythemia is rare, reticulocytosis. Occasionally there are nucleated cells. Leukocytes normal by 40%, elevated by 40%, decreased by 20%. Increase in basophils. Sometimes nucleated cells. Elevated leukocyte alkaline phosphatase.

Elevated uric acid. Moderate hyperbilirubinemia. Normal or decreased platelets. Giant platelets.

Medulogram. Increase in bone consistency.

Myelofibrosis. Marked hypoplasia of cellular systems.

Splenic puncture. myeloid hyperplasia.

bone radiology. Typical changes are observed.

Syndromogenesis and etiology

There is hyperplasia of the spleen due to myeloid metaplasia.

It can also be seen in the liver and lymph nodes.

The etiology is unknown.

Literature

Stephens, D.J. and J.F. Bredek: "A leukemic myelosis with osteosclerosis." Ann. Int. Med., 6:1087-1096, 1933.

MOSCHCOWITZ SYNDROME

Synonymy

S. of thrombotic thrombocytopenic purpura. S. of thrombotic thrombohemolytic purpura. S. of thrombocytic angiothrombosis.

S. of microangiopathic hemolytic anemia. S. de Baehr-Schriffrin.

Syndromography

Clínic

It occurs between 10 and 40 years of age, with a predominance in women. Neurological symptoms fluctuate: headache, paresis, syncope, aphasia, dysarthria, visual changes, coma.

Hemorrhagic manifestations are constant (purpura, retinal hemorrhages, melena, hematemesis, hematuria).

Physical examination. Pallor, petechiae, ecchymosis, jaundice (not constant) are detected. Moderate lymphadenopathy, hepatosplenomegaly.

Paraclinical examinations

Blood. Thrombocytopenia, occasionally very severe anemia. Reticulocytosis. Fragmented cells. Hyperbilirubinemia. Negative Coombs test. Increase in leukocytes (leukemoid reaction). High urea.

Medulogram. Increase in immature megakaryocytes and myeloid and erythroid hyperplasia.

Urine. Proteinuria, hematuria and casts.

Syndromogenesis and etiology

This syndrome is caused by intravascular thrombosis of unknown etiology, although an autoimmune mechanism has been described.

Literature

Amorosi, E.L. and J.E. Ultmann: "Thrombotic thrombocytopenic purpura; report of 16 cases and review of the literature." Medi- cine, 45: 1 39, 1966.

Karpatkin, S.; M. Folino and R. Winchester: "Hereditary autoimmune thronbocytopenic purpura. an immunological and genetic study." Ann. Intern. Med., 94:781-782, 1981.

Moschcowitz, E.: "Acute febrile pleiochromic anemia with hyaline thrombosis of terminal arterioles and capillaries; an undescribed disease." Arch. Int. Med. 36: 89-93, 1925.

MOSSE SYNDROME

Synonymy

S. of polycythemia with liver cirrhosis.

Syndromography

Clinic

According to Mosse, polycythemic symptoms appear before those of cirrhosis.

Paraclinical examinations

Blood. Polycythemia. Alteration of liver function tests. Low fibrinogen.

Medulogram. Hyperplasia of the erythropoietic system.

Liver biopsy. Cirrhosis.

Syndromogenesis and etiology

The etiology has not been established. It is debated whether it is an autonomous syndrome or a simple coincidence.

Literature

Mosse, M.: "Ueber Poleythemie mit Uroblinikterus und Milztumor" Deut. Med. Wscher., 33:2175-2176. 1907.

BENIGN FAMILY CHRONIC NEUTROPENIA SYNDROME

Synonymy

S. de Gansslen.

Syndromography

Clinic

A family history is collected. Symptoms vary from an asymptomatic form to a tendency toward chronic recurrent infections. The severe generalized form is occasional.

Physical examination. Evidence of periodontal disease is found. There is no splenomegaly.

Paraclinical examinations

Blood. Leukocytes are on the border of the lowest range; There is absolute neutropenia and relative lymphocytosis, monocytosis, and a very irregular increase in eosinophils.

Medulogram. Normal erythropoietic and megakaryopoietic series. Marked reduction of the most mature elements, increase in lymphocytes and monocytes.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Cutting, H.0. and J.E. Lang: "Familial benign chronic neutropenia." Ann. Int. Med., 61:876-887, 1964.

Gansslen, M.: "Konstitutionelle familiale Leukopenia (Neutropenie)" . Klin. Wschr., 20:922-925, 1941.

HEREDITARY HEMOLYTIC OVALOCYTOSIS SYNDROME

Syndromography

Clínic

This syndrome affects both sexes and from childhood. There are varying degrees of hemolysis occasionally associated with splenomegaly. Ulcerations in lower limbs. Maxillofacial-dental bone anomalies.

Paraclinical examinations

Blood. Increase in osmotic fragility. Typical oval-shaped erythrocytes and increased autohemolysis are observed. Decreased erythrocyte survival.

Syndromogenesis and etiology

Of genetic etiology. It is transmitted by autosomal dominant inheritance.

The basic defect is not known.

Literature

Cutting, H. O.; W.J. McHugh; R.G. Conrad, et al.: "Autosomal dominant hemolytic anemia characterized by ovalocytosis." Amer. J. Med., 39:21-34, 1965.

PARAHEMOPHILIA SYNDROME

Synonymy

S. of deficiency of the factor V. S. of Owren (I).

Syndromography

Clinic

Parahemophilia affects both sexes. It is very variable in severity and way of settling. The symptoms are similar to those of hemophilia except hemarthroses, which do not occur in this syndrome. There is menorrhagia, hemorrhage a few days after delivery. Mucous membrane hemorrhages.

Paraclinical examinations

Blood. Prolonged prothrombin time that is amended with fresh plasma. Absence of coagulation factors that are dependent on vitamin K, which are not replaced with aged or oxalated plasma. Abnormal thromboplastin generation test. Altered prothrombin consumption.

Factor V deficiency.

Syndromogenesis and etiology

This syndrome is due to factor V deficiency, which determines a prolongation of prothrombin and coagulation time.

Of genetic etiology, it is transmitted by autosomal recessive inheritance.

Bibliography

Owren, P.A.: "The coagulation of blood: investigations of a new clotting factor." Acta Med. Scandinav., 128:Suppl. 194:1-327, 1947.

PELGER-HUET SYNDROME

Synonymy

S. de Pelger.

Syndromography

Clinic

This syndrome is found in Germany and Holland (1:1,000) and also in the United States (1:10,000). He is asymptomatic. Occasionally it is associated with other anomalies.

Paraclinical examinations

Blood. Neutrophils have an eccentric and frequently fragmented nucleus. Normal maturation of the cytoplasm. Chromatin condensation in lymphocytes, monocytes, megakaryocytes and

Syndromogenesis and etiology

Of genetic etiology. It is transmitted by autosomal dominant inheritance. Sometimes just the trait is inherited. It is interesting to remember that some of the leukocyte abnormalities can be observed in cases of leukemia, Fanconi anemia and during treatment with myelotoxic agents. This is called Pelgeroid malformation.

Literature

Huet, G.J.: "Over een familiare anomalie der leucocyten". Mschr. Kindergeneseek, 1:173-181, 1932. abstr. Nederl. tijdschr. v. Geneesk., 75:5965-5959. 1931.

Miale, J.B.: Laboratory Medicine, Hematology. St. Louis, Mosby, 1967.

Pelger, K.: “Demonstratie van een paar zeldzaam voorkomende types van bloedichaampjes en bespreking der patients.” Nederl. T. Geneesk., 72:1178, 1928.

PIRINGER KUCHINCA SYNDROME

Synonymy

S. of Piringer Kuchinca type lymphadenitis.

Syndromography

Clinic

This syndrome was described by Alexandra Piringer Kuchinca (Austrian pathologist) in 1952.

It is characterized by recurrent tonsillitis with rheumatoid joint manifestations accompanied by slightly painful lymph node swellings that can reach the size of a walnut in the neck and nape region. There are no general symptoms or fever.

Chronic infection of the nose and throat. The prognosis is favorable with regression of clinical manifestations in a

month.

Paraclinical examinations

Blood. Marked increase in beta and gamma globulins.

Normal total proteins.

Node biopsy. Epithelioid-type reticular proliferation in the stroma and cords. No necrosis or hyalinosis are observed. A characteristic argyrophilic granulation is seen in the protoplasm of the epithelioid cells.

Syndromogenesis and etiology

It is probably a tissue reaction to a large number of etiological factors. Toxoplasmosis, which is found in 95% of cases, seems to play an important role. It has also been considered a chronic expression of Pfeiffer glandular fever.

The possibility of its transformation into Hodgkin or Besnier-Boeck syndrome is discussed.

Literature

Piringer Kuchinca, A.: "Bigenartige mikroskopiche Befunde am Lynphknoten der oberen Körperhälfte." Verb. Dtsh. Path. Ges., 36:522-535, 1952.

TUMORS-ASSOCIATED POLYCYTHEMIA SYNDROME

Syndromography

Clinic

As a cause of this syndrome, the association of an increase in erythrocytes without leukocytosis, thrombocytosis, or splenomegaly with a variety of neoplasms has been reported. This syndrome has occurred in pituitary tumors, vascular, infratentorial, and renal tumors (hypernephroma, renal cysts, polycystic kidneys, Wilm's tumor). Hepatoma, pheochromocytoma, uterine fibromyoma, undifferentiated carcinoma of the lung, congenital adrenal hyperplasia with polycythemia (see).

Syndromogenesis and etiology

The secretion of an erythropoiesis-stimulating substance that would be secreted by different tumors is noted. Removal of the tumors eliminates polycythemia.

Literature

Castex, Mariano R. (1929) quoted by Guillain: G., P. Lechelle and R. Garcin: " La polyglobulic avec ou sans erythrose, de certain syndromes hypophyso-tuberiens." Ann. of Med., 31:100-114, 1932.

ALLOGENEOUS POST-TRANSFUSION SYNDROME

Synonymy

S. of bone marrow transfusion. S. of leukocyte transfusion.

Syndromography

Clínic

Allogeneic posttransfusion syndrome occurs in patients who, after bone marrow irradiation, received a transfusion of allogeneic or leukocyte bone marrow. Symptoms begin 5-10 days after transfusion with fever, asthenia, anorexia, nausea, vomiting, diarrhea and severe weight loss.

Physical examination. Exfoliative erythroderma is detected.

Occasionally jaundice and hepatomegaly, and tran-

history.

Paraclinical examinations

Blood. Transient lymphocytosis followed by intense or moderate lymphocytopenia. There may be anemia. Decrease in immunoglobulins G, My A.

Blood culture. You can find bacteria, viruses,

fungus.

Syndromogenesis and etiology

The syndrome is considered due to a graft-versus-host reaction.

Literature

Mathe, G; L. Schwarzenberg: M.J. de Vries, et al.: "Les divers aspects du syndrome secondaire compliquant les transfusions allogenes de moelle osseuse ou de leukocytes chez des sujects atteints d'hemopathies malignes." Europ. J. Canc., 1:75-113, 1965.

WERLHOF PURPLE SYNDROME

Synonymy

S. of immunological thrombocytopenic purpura. S. of acute idiopathic thrombocytopenic purpura. S. of pseudohemophilia. S. Werlhof. S. of hemorrhagic purpura.

Syndromography

Clinic

This syndrome predominates in children (85%) under 8 years of age. It affects both sexes equally. It is generally preceded, within one to two weeks, by a viral infectious process.

Acute purpuric manifestations occur on the skin and mucous membranes, including petechiae and ecchymosis. In adults the onset may be gradual.

In acute cases there is severe bleeding in the mouth, anus, bladder or genitals. The bleeding mucosa shows no signs of inflammation or ulcerations.

In the chronic form, this syndrome presents poussées without great intensity. There is moderate splenomegaly, but there is an absence of joint or abdominal manifestations.

Paraclinical examinations

Blood. Anemia, thrombocytopenia less than 30,000, with alterations in platelets (giant, agglutinated or immature with few granules). Shortened platelet survival time. Very long bleeding time.

Normal clotting time but clot retraction is defective or absent. Positive prothrombin consumption test.

Antiplatelet factors have been demonstrated.

Medulogram. Increase in megakaryocytes, with qualitative alterations (giants).

Syndromogenesis and etiology

Heterogeneous condition of multiple etiology. In its pathogenesis, the action of antibodies generated in the spleen is invoked, during viral infections, or secondary to medications:
quinine, digitoxin, chlorothiazide, tolbutamide, barbiturates, meprobamate, sulfas, antibiotics, salicylates, etc., which were previously well tolerated.

The binding of the virus and antibody on the platelet surface is invoked, with or without complement activation.

There is possible interference with the function of T-suppressor lymphocytes.

The etiology is unknown.

Literature

Frank, E.: “Die essentielle thrombopenie.” Berl. Klin. Wchr., 52:454-458, 1915.

MeClure, P.D.: Idiopathic thrombocytopenic purpura in children: diagnosis and management. Pediatrics, 55:68-74, 1975.

Werlhof, P.G.: Disquisitio medica et philologica ed variolis et anthracibus. Brunswick, 1735.

PURPURA FACTICIA SYNDROME

Synonymy

S. of devil's pinch.

Syndromography

Clinic

In this syndrome there are manifestations of painful ecchymosis.

recurring roses.

Paraclinical examinations

If protection with plastic is placed over the area, ecchymosis does not appear.

Coagulogram. Normal.

Syndromogenesis and etiology

It is a syndrome in which ecchymoses are self-induced consciously or unconsciously.

Literature

Davidson, E.: "Factitious purpura presenting as autoerythrocyte sensitization." Brit. Med. J. 1:104, 1964.

HEREDITARY FAMILY PURpura SYNDROME

Syndromography

Clinic

The symptoms of this syndrome predominate in women and are frequently associated with rheumatic fever.

Syndromogenesis and etiology

It is debated whether it corresponds to a family hereditary pattern. The etiology is unknown.

Purpura fulminans syndrome

Synonymy

S. de Gimard. S. of Scheldon. S. of hemorrhagic gangrenous purpura. S. of gangrenous purpura.

Syndromography

Clínica

This syndrome is more common in children, appears in association with various infections (viral, streptococcus) and also with pregnancy.

It begins suddenly: fever, prostration, ecchymoses spread on the skin, it does not affect the mucous membranes. Gangrene can occur quickly.

Paraclinical examinations

Blood. Some cases may present factor V deficiency, excess antithrombin, increased fibrinogen.

Syndromogenesis and etiology

There may be an allergic mechanism similar to the Schwartzmann phenomenon.

The etiology is unknown.

Literature

De Gimard, M.: "Purpura hemorrhagique primitif au purpura infectieux primitif", Paris, 1844 (Thesis) quoted by Wintrobe, M.M.: Clinical Hematology ed. 6, Philadelphia, Lea & Febiger, 1967.

MECHANICAL PURPLE SYNDROME

Syndromography

Clinic

In this syndrome, purpuric manifestations occur, usually in the lower limbs in patients who have an acute and rapid increase in capillary pressure in the course of convulsions, cough, use of tourniquets, chronic increase in pressure (orthostatic position).

Paraclinical examinations

Platelet count.

Clotting and bleeding time. Normal.

Syndromogenesis and etiology

A congenital or acquired factor is invoked as responsible for capillary weakness, especially in orthostatic type.

POST-TRANSFUSIONAL PURpura SYNDROME

Synonymy

Shulman S.

Syndromography

Clínica

It is a rare reaction reported only in middle age, in women. One week after repeated transfusions, severe purpura occurs.

Paraclinical examinations

Blood. Severe thrombocytopenia with antiplatelet antibodies. Positive platelet agglutination tests.

Syndromogenesis and etiology

It is possibly due to a cross reaction with destruction of the patient's platelets.

Literature

Morrison, F.S. and P.L. Mollison: "Purple post-transfusion." New Eng. J. Med., 275:243-248, 1966.

Van Loghem, J.J.; H. Dorfmeijer and M. Van der Hart: "Serological and genetic studies on platelet antigen (Zw.)". Vox Sang. 4:161-169. 1959

SENIL PURPURA SYNDROME

Synonymy

S. of Bateman. S. of cachectic purple.

Syndromography

Clinic

Senile purpura occurs in elderly and cachectic individuals. Small petechiae and hematomas, particularly on the forearms and back of the hands, 1-4 cm in diameter, slowly reabsorbed, leaving pigmentation.

Syndromogenesis and etiology

There is an increase in vascular fragility.

SIMPLE PURpura SYNDROME

Syndromography

Clínic

Symptoms predominate in women. Moderate purpura often appears during menstrual periods.

Paraclinical examinations

Blood. Normal platelet count, normal coagulation factors.

Loop test. Sometimes it shows abnormalities.

Syndromogenesis and etiology

Genetic etiology with dominant inheritance is suggested, but without male-to-male transmission. The basic defect is unknown.

Literature

Davis, E.: “Hereditary familial purpura simplex: review of 27 families.” Lancet, I:145-146. 1941.

CHRONIC IDIOPATHIC THROMBOCYTOPENIC PURpura SYNDROME

Syndromography

Clínic

This syndrome predominates in women and begins in adulthood, insidiously. It is accompanied by heavy menstruation and moderate bleeding. The spleen may be normal or there may be slight splenomegaly.

Paraclinical examinations

Blood. Moderate anemia due to hemorrhages. Normal leukogram. Moderate thrombocytopenia (40,000-80,000). Shortened platelet survival. Antiplatelet factors are detected in sera (IgG antibodies).

Syndromogenesis and etiology

It is possibly a syndrome caused by an autoimmune mechanism. It may appear as a complication of other diseases associated with an immune disorder (lupus erythematosus, lymphoproliferative syndromes, such as Hodgkin's disease). Thrombocytopenia is the result of antibodies reacting with platelet antigens.

The etiology is unknown.

Literature

Baldini, M.: "Idiopathic thrombocytopenic purpura." New Eng. J. Med., 274:1 245-1 251:1 360-1 367, 1966.

Moake, J.L.: "Common bleeding problems." Clinical Symposia. Ciba. v. 35 M3 1983.

RECURRENT IDIOPATHIC THROMBOCYTOPENIC PURpura SYNDROME

Syndromography

Clínic

(See chronic idiopathic thrombocytopenic purpura.)

This form is characterized by recurrent episodes with complete hematological and clinical remissions.

Paraclinical examinations

Blood. Regression to normal of the number of platelets and their survival in remissions. During crises there is thrombocytopenia.

Literature

Baldini, M.: "Idiopathic thrombocytopenie purpura." New Eng. J. Med. 274:1 245-1 251: 1 360-1 367, 1966.

Frank, E.: “Die essentielle Thrombopenie.” Berl. Klin. Wchr., 52:454-458, 1915.

Werlhof, P.G.: Disquisito medica et philologica ed variolis et anthracibus. Brunswick, 1735.

RIETTI-GREPPI SYNDROME

Synonymy

S. of microelliptopoikilocytic anemia. S. of thalassemia minor. Heterozygous beta-thalassemia S.

Syndromography

Clínic

This syndrome is often asymptomatic and the presence of iron-resistant hypochromic anemia is a casual finding. Hemolytic jaundice and decreased osmoic fragility.
Splenomegaly, discrete in half of the cases. Osteopathy sometimes, minimal, with "brush skull".

Paraclinical examinations

Hemogram. It shows the presence of anemia, mild 10-11 g of hemoglobin/100 mL; microcytic, with anisopoikilocytosis, basophilic stippling and increased globular resistance. Hemoglobin electrophoresis shows an increase in A2 (4-6%): in half of the cases there is an accumulation of Hb.F (1-3%).

Medulogram. Hyperplastic erythroid marrow, with normoblasts.

Serum iron. Diminished.

indirect bilirubin. Increased.

Osmotic fragility. Discretely increased.

Syndromogenesis and etiology

The syndrome is caused by an abnormality in globin synthesis with decreased hemoglobin synthesis.

It is the heterozygous form of beta-thalassemia, with an increase in hemoglobin A2 and sometimes in Hb Fo "Lepore"

The most important thing is to inform heterozygous couples that 25% of their children will develop a fatal form (Cooley syndrome) .

Literature

Greppi, E.: "Itero emolitico familiare con increase della resistance dei globuli." Minerva Med., (part 2) 8:1-11, 1928.

Marks, P.A.: "Thalassemia syndromes. Biochemical, genetic and clinical aspects." New Eng. J. Med., 275:1363-1369, 1966.

Micheli, E: "Le splenomegalic emolitiche." Att. 350. Cong. Soc. Italian. Med. Int. 1929.

Rietti, F.: "Sugli itteri emolitici primitivi." Atti. Acc. Sci. Med., Nat. Ferrara, 1925.

DNA SENSITIVITY SYNDROME

Syndromography

Clinic

In this syndrome, painful spots appear on the limbs that precede the appearance of hives or nodules. Within 2-48 hours, welts or red nodules are found within hematomas that spread circumferentially with a diameter of 10-12 cm, sometimes from the middle of the hip to the ankle or from the elbow to the tips of the fingers. These lesions appear at intervals of 2-4 weeks.

Paraclinical examinations

The injection of DNA preparations into the skin of the members reproduces the specific lesions. Injection into the skin of the trunk gives a negative test.

Coagulogram. Normal.

Syndromogenesis and etiology

There is self-sensitization to DNA limited to the members.

The etiology is unknown.

Literature

Levin, M.B. and H. Pinkus: "Autosensitivity to deoxyribonucleic acid (DNA). Report of a case with inflammatory skin lesions controlled by chloroquine." New English J. Med., 264:533-537, 1961.
Little, A.S. and H.E. Bell: "Painful subcutaneous hemorrhages of the extremities with unusual reactions to injected deoxyribonucleic acid." Ann. Int. Med., 60:886-891, 1964.

SILVESTRONI-BIANCO SYNDROME

Synonymy

S. of microsickling. S. of sickle cell thalassemia. Thalassemia minimal S. S. beta-thalassemic.

Syndromography

Clínic

There is moderate or severe anemia, including manifestations of Herrick syndrome and thalassemic syndrome. Abdominal pain, splenomegaly.

Paraclinical examinations

Blood. Anemia. Findings of sickle cell anemia and thalassemia.

Hemoglobin electrophoresis. It shows 60-80% Hb S and 20% Hb F.

Syndromogenesis and etiology

This syndrome is a genetic combination of both sickle cell anemia and thalassemia, each inherited from one parent, rarely from both.

Literature

Silvestroni, E. and I. Bianco: "Ricerche cliniche, genetiche et dermatologiche sui malati di anemia microcytica constituzionable e di morbo di Cooley." Haematologica, 3:135-190, 1948.
Silvestroni, E. and I. Bianco: "Genetic aspects of sickle cell anemia and microsickling disease." Blood, 7:429-435, 1952.

COOLEY THALASSEMIA SYNDROME

Synonymy

S. of erythroblastic anemia. S. of Mediterranean anemia. S. of hereditary leptocytosis. S. of thalassemia major.

Syndromography

Clinic

It appears in both sexes. It begins insidiously at 3-6 months of age. There is severe weakness, mongoloid facies, prominent cheekbones, thickening of the gums, prominent teeth and sometimes epicanthus, torricephaly, abdominal distention, hepatomegaly and marked splenomegaly. Recurrent febrile episodes, cardiomegaly, hemorrhagic manifestations.

Intense pallor with subicteric tinge in the conjunctivae.
Cardiac dilation with anemic murmur. There is often growth retardation.

Paraclinical examinations

Blood. There is severe anemia. Marked anisopoikilocytosis.
Cellular distortion and deformity. Marked reticulocytosis.
Red blood cells in "target". Marked increase in fragility
Osmotic, Generally leukocytosis with immature forms.
Hyperbilirubinemia. Elevated serum iron. Decreased erythrocyte survival time.

Urine. Increased urobilirubinogen, urobilin and aminoaciduria.

Stool. Increased coproporphyrin.

Radiology. Skull, radial striae of the diploe with brush skull appearance. Thickening of the vault with thinned cortex. Long bones, decreased marrow density and thickening of the cortex. Limbs, thin cortex, expanded spinal canal, osteoporosis, sometimes spontaneous fractures.
Chest, cardiac dilation, widening of the ribs, osteoporosis.

Medulogram. Intense hyperplasia of the red blood cell series in all its maturation stages. There may be a crisis of spinal aplasia due to exhaustion, sometimes preceded by infection or ingestion of some medications.

Syndromogenesis and etiology

The basic defect consists of the failure of the synthesis of a polypeptide chain, by the ribosomes of the cytoplasm of the medullary normoblasts, the globin being defective, it does not bind the iron, which accumulates in the cell and the red blood cells are poor in hemoglobin, hypochromatic and short-lived. In thalassemias, one of the amino acids of one of the globin chains is changed, which can occur in the alpha, beta or gamma chain. In Cooley's thalassemia, the beta chain is affected.

The etiology is genetic and follows Mendelian laws. This syndrome is seen in homozygotes. The prognosis is poor, it is always fatal, with survival of around 20 years in well-treated cases; They are threatened by sepsis and myocardial siderosis.

Literature

Cooley, T.B. and P. Lee: "Series of cases of splenomegaly in children with anemia and peculiar bone change." Trans. Am.
Pediat. Soc. 37:29, 1925.
Marks, P.A.: "Thalassemia syndromes. Biochemical, genetic, and clinical aspects." New Eng. J. Med. 275: 1363-1369, 1966.

HEMORRHAGIC THROMBOCYTHEMIA SYNDROME

Synonymy

S. of idiopathic hemorrhagic thrombocythemia. S. de Mortensen. Epstein's S.

Syndromnography

Clinic

It affects both sexes equally and begins in the third decade or somewhat later. It is characterized by spontaneous hemorrhagic manifestations of variable intensity, hemoptysis, melena, menorrhagia, etc.

There are post-traumatic or post-surgery hemorrhages. Thrombosis may occur in the splenic vein and in the deep and superficial veins of the legs. Splenomegaly.

Paraclinical examinations

Blood. Anemia, occasionally. Erythrocytosis. Leukocytes are between 10,000-30,000, occasionally above 60,000. Increase in platelet numbers between 1-6 million. In some cases there is a deficiency of factors II, V and VII.

Medulogram. Hyperplasia of the three systems, with a marked increase in megakaryocytes, some abnormalities can be observed.

Syndromogenesis and etiology

This is probably a thrombocytic variant of Vaquez-Osler syndrome.

The etiology is unknown.

Literature

Epstein, E. and A. Goedel: “Hamorrhagische Thrombocythamie bei vasculare. ” Schrumpfmilz. Virch.Arch., 292:233-248, 1934.
Gunz, FW.: "Hemorrhagic thrombocythemia; a critical review." Blood, 15:706-723, 1960.

PRIMARY HYPERSPLENIC THROMBOCYTOPENIA SYNDROME

Syndromography

Clinic

It affects both sexes, and can occur at all ages. There are varying degrees of hemorrhagic manifestations. Splenomegaly.

Paraclinical examinations

Blood. Anemia, neutropenia.

Medulogram. Hyperplasia of the erythropoietic, granulopoietic and megakaryopoietic systems.

Syndromogenesis and etiology

There is dysfunction of the spleen with trapping and destruction of

The etiology is unknown.

Literature

Cooney, D.P. and B.A. Smith: "The pathophysiology of hypersplenic thrombocytopenia." Arch. Int. Med. 121:332-337, 1968.

AUTOSOMAL DOMINANT THROMBOPATHIC THROMBOCYTOPENIA SYNDROME

Synonymy

S. of congenital hemorrhagic thrombocytic dystrophy. S. of hereditary thrombopathic thrombocytopenia. S. de Bernard-Soulier.

Syndromography

Clinic

It occurs in both sexes, and is manifested by epistaxis, menorrhagia, and spontaneous hemorrhages early in life. Deep (intramuscular) hemorrhages are rare.

In severe cases there are intracerebral and intraocular hemorrhages. Occasionally petechiae occur.

Paraclinical examinations

Blood. Normal clotting time. Prolonged bleeding time. Thrombocytopenia. Decreased prothrombin consumption. Prolonged clot retraction.

Loop test. Positive.

Syndromogenesis and etiology

Of genetic etiology. It is transmitted in an autosomal dominant manner.

The basic defect is not clarified. Cases associated with platelet autoantibodies have been reported.

Literature

Bernard, J. and J.P. Soulier: Sur une nouvelle varieté de dy strophie thrombocytaire hemorragipare congenitale". Bull. et Mem. Soc. Med. Hop. Paris, 64:969-974, Oct. 22-29, 1948.
Quick, A.J.: Hemorrhagic Diseases and Thrombosis. ed. 2, Philadelphia, Lea & Febiger, 1966.
Najean, Y and T. Lecompte: "Genetic thrombocytopenia with autosomal dominant transmission: a review of 54 cases." Brit. J. Haemat., 74:203-208, 1990.

SEX-LINKED RECESIVE THROMBOCYTOPEN SYNDROME

Syndromography

Clinic

It only occurs in men. Women have minor thrombocytopenia and moderate bleeding. Severe hemorrhages are seen in men.

Syndromogenesis and etiology

Thrombocytopenia has been reported associated with reduced platelet volume. It is a genetic defect linked to the recessive expression chromosome.

Literature

Bernard, J. and J.P. Soulier: "Sur une nouvelle varieté de dystrophie thrombocytaire hemorragipare congenitale." Bull. et Mem. Soc. Med. Hop. Paris, 64:969-974, Oct. 22-29, 1948.
Quick, A.J.: Hemorrhagic Diseases and Thrombosis. ed. 2, Philadelphia, Lea & Febiger, 1966.

VAQUEZ-OSLER SYNDROME

Synonymy

S. of polycythemia vera. S. of splenomegalic polycythemia. S. of Vaquez. S. de Osler. S. of polycythemia rubra. S. of myelopathic polycythemia. S. of megalosplenic erythrocytosis. Cryptogenetic polycythemia S.

Syndromography

Clinic

Discreet predominance in men. It occurs in middle or older age. Occasionally in children. It installs insidiously. There is headache, syncope, visual disturbances, ringing in the ears, dyspnea on exertion, itching after bathing, epistaxis, gingivorgia, paresthesia.

Physical examination. Plethoric facies, color changes in limbs. Ecchymoses of various sizes. Purple in 8% of cases. Conjunctival congestion. Engorgement of veins. Clubbing fingers. Hepatomegaly. Hard and smooth splenomegaly.

Paraclinical examinations

Blood. Red blood cells between 7-10 million. Hemoglobin above 18-24 g%. Leukocytes above 10,000. Phosphatase

alkaline above normal values. Platelets increased to more than 3-6 million. Increase in viscosity. Increased volume. Normal or elevated uric acid.

Marrow biopsy. Hypercellular marrow with hyperplasia of the three systems.

Urine. Normally or rarely proteinuria.

Syndromogenesis and etiology

It is part of the myeloproliferative syndromes.

The etiology is unknown although familial cases have been reported and a genetic etiology is suggested.

Literature

Osler, W.: Chronic cyanosis with polycythemia and enlarged spleen a new clinical entity. Amer. J. Med. Sci., 126:187-201, 1903.

Vaquez, H.: "Sur une forme spéciale de cyanose sáccompagnant d' hyperglobulie excessive et persistent." Compt. Yield. Med. Par., 44:384-388, 1892.

WALDENSTRÖM SYNDROME I

Synonymy

Hyperglobulinemic purpura S. S. of dysproteinemic primitive

Syndromography

Clinic

Chronic micromacular purpura, especially on the legs, with hyperpigmented spots as a result of blood extravasation. No hemorrhages are observed in the mucous membranes.

Relatively benign chronic condition, recurrent depending on the underlying disease. Rumpel test- -Leed positive.

Paraclinical examinations

Blood. Normal bleeding and clotting time. Normal thrombocythemia. Increased erythrocyte sedimentation rate.

Marked increase in the gamma fraction of serum globulins.

Syndromogenesis and etiology

The possibility of dysproteinemia or paraproteinemia in the course of an underlying disease, with consequent capillary damage, arises.

Literature

Waldenström, J.: «Zwei interesting Syndrome mit Hyperglobulinämia purpura hyperglobulinemica und Makroglobulinämie. Schweis. Med. Wschr., 78:927, 1948.

WALDENSTRÖM II SYNDROME

Synonymy

S. of idiopathic macroglobulinemia. S. of primary macroglobulinemia.

Syndromography

Clínica

It occurs in middle age. Both sexes are affected. The patient reports asthenia, weight loss, repeated infections, visual disorders, and hemorrhagic manifestations.

physical examination Dyspnea, paleness, microlymphadenopathy, variable hepatosplenomegaly. Fundus: sausage-shaped veins are observed (paraproteinemic fundus).

Acrocyanosis.

Evolution almost always lethal.

Paraclinical examinations

Blood. Anemia, leukopenia and thrombocytopenia. Rouleaux phenomenon. SIA and formalin-gel tests, positive.

Monoclonal hypergamma (Component "M").

Macroglobulin in ultracentrifugation. Decrease in the rest of the immunoglobulins. Accelerated erythrocyte sedimentation.

Medulogram. Increase in plasma cells and lymphocytes adults.

Syndromogenesis and etiology

This syndrome can be primary, associated with lymphocytic proliferation; or secondary, associated with lymphomas, collagenopathies, chronic infections.

Literature

O'Reilly, R.A. and M.R. MacKenzie: "Primary macrocryogel-globulinemia. Remission with adrenal corticosteroid therapy". Arch. Int. Med., 120:234-238, 1967.

Waldenström, J.: "Incipient myelomatosis or *essential' hyperglobulinemia with fibrinogenopenia- a new syndrome?". Acta Medica, Scandinav., 117:216-247, 1944.

WIDAL-RAVAUT SYNDROME

Synonymy

S. de Jaffe-Gottfried-Bradley. S. of non-spherocytic hemolytic anemia.

Syndromography

Clínic

The syndrome affects both sexes. It settles in childhood.

It is characterized by chronic hemolysis. It also has different stages of onset, but generally presents during infancy and childhood.

Different deficiency syndromes have been recognized: glucose-6-phosphatedehydrogenase, pyruvatokinase, glutathione reductase, glutathione, triosephosphateisomerase, formiminotransferase, Jaffe-Gottfried-Bradley, Bassen-Kornzweig, and others.

These terms are considered a group of syndromes with varying degrees of hemolytic anemia, whose characteristic is that they do not benefit or do little after splenectomy.

Paraclinical examinations

Blood. Moderate anemia, reticulocytosis. Moderate hyperbilirubinemia. Decreased osmotic fragility, slightly increased autohemolysis that is corrected by the addition of glucose. Normal erythrocyte enzymes.

There is absence of spherocytosis or increased osmotic fragility.

Dacie described 2 types:

Type I. With oval macrocytosis. Autohemolysis that improves with the addition of glucose.

Type II. Rounded macrocytes, autohemolysis and greater potassium loss than the normal erythrocyte, which is not modified by the addition of glucose.

Syndromogenesis and etiology

It is due to an alteration of the erythrocyte phospholipids or membrane abnormality.

It is transmitted by autosomal recessive inheritance.

The basic defect is not known.

Literature

Dacie, J.V.; P.L. Mollison; N. Richardson, et al.: "Atypical congenital haemolytic anaemia." Quart. J. Med., 22:79-98, 1953.

Jaffe, E.R: E.L. Gottfried and T.B. Jr. Bradley: "Hereditary nonspherocytic hemolytic disease associated with altered phospholipid composition of erythrocytes." J. Clin. Invest. 45:1027, 1966.

Jaffe, E.R. and E.L. Gottfried: “Hereditary nonspherocytic hemolytic disease associated with an altered phospholipid composition of the erythrocytes.” J. Clin. Invest., 47:1375- -1388, 1968.

Widal, F. and P. Ravaut: "Ictére chronique acholurique congenital chez un homme de vingt-neuf ans." Bull. et Mem. Soc. Méd. Hop. Paris, 3 f., 19:984-991, 1902.

WILLEBRAND JÜRGENS SYNDROME

Synonymy

S. von Willebrand. S. of constitutional thrombopathy. S. of pseudohemophilia. S. of vascular hemophilia. S. of angiohemophilia.

Syndromography

Clinic

It affects both sexes. It usually appears early in childhood. There are hemorrhagic manifestations varying in intensity. Tendency to easy bleeding, ecchymosis, epistaxis, gingivorrhagia, urinary and digestive hemorrhagic manifestations. Prolonged bleeding after trauma or surgery.

Paraclinical examinations

Blood. A deficiency of factor VII, Von Willebrand and reduction of Factor VIIIc is found. Normal loop test.

Prolonged bleeding time. Abnormal partial thromboplastin time and clotting time. Abnormal prothrombin consumption.

Radioimmunoassay. Decrease in factor VIII vw.

One-dimensional and two-dimensional immunoelectrophoresis. Normal platelet count.

Syndromogenesis and etiology

It is an autosomal dominant disease with variable penetrance and expressivity.

It occurs as a result of a decrease in the release of von Willebrand factor VII by endothelial cells (type I).

Defect in type II polymerization (A) or abnormal synthesis of polymers II (B). All of which reduces platelet adhesiveness to the damaged endothelium.

Literature

Moake, J.L.: "Common Bleeding Problems." Clinical Symposia. Ciba. v. 35 No.3. 1983.

RELATED HEMOLYMPHOPOIETIC SYNDROMES

Syndromes with megaloblastic anemia

Addison-Biermer.

Hereditary orotic aciduria.

Congenital intrinsic factor deficiency.

Lichtheim.

Malabsorption-small intestine diverticulosis-anemia macrocytic.

Hereditary intestinal malabsorption of Vitamin B12.

Non-tropical sprue.

Tropical sprue.

Syndromes with defibrination

Acquired hypofibrinogenemia syndromes will be considered since congenital hypofibrinogenemia or afibrinogenemia syndromes only have one cause.

The conditions that cause this syndrome give rise to severe bleeding and are the following:

1. High hematocrit and inadequate fibrinogen to form a good clot:

Cyanotic congenital heart disease.

Vaquez-Osler.

2. Decreased fibrinogen synthesis:

Amyloidosis.

Liver diseases (rare).

3. Excessive use that causes a deficiency:

Septic abortion.

Abrupt placenta (placental abruption).

Extracorporeal circulation.

Amniotic fluid embolism.

Dead fetus.

Giant cavernous hemangioma.

Hypofibrinogenemia in digestive neoplasms.

Moschcowitz.

Lightning purple.

Incompatible blood transfusion.

Massive venous thrombosis.

Waterhouse-Friderichsen.

4. Fibrinolysis:

Primary (hyperplasminemia):

Inhibitor deficiency (liver diseases).

Iatrogenic (administration of plasminogen activator).

Endogenous release (surgery in lung, prostate, brain neoplasms).

Other proteolytic enzymes (leukemias).

Secondary (association of defibrination and fibrinolysis):

Amniotic fluid embolism.

Dead fetus.

Very severe infections.

Kasabach-Merritt.

Leukemias.

Lymphomas.

Placenta previa.

Lightning purple.

Waterhouse-Friderichsen.

Syndromes with hemoglobinopathies

Hemoglobin C.

Hemoglobin D.

Hemoglobin E.

Hemoglobin H.

Herrick.

Jaksch-Hayem-Luzet.

Rietti-Greppi-Micheli.

Silvestroni-Bianco.

Thalassemia.

Hemolytic syndromes

Anemia with Heinz bodies in the newborn.

Hemolytic anemia.

Acquired idiopathic hemolytic anemia.

Anemia-pigmenturia-Bassen inclusion bodies-

-Kornzweig.

Formiminotransferase deficiency.

Glucose-6-phosphate dehydrogenase deficiency.

Glutathione deficiency.

Glutathione reductase deficiency.

Triose-isomerase deficiency.

Doan-Wright.

Evan.

Favism.

Black water fever.

Haff.

Paroxysmal hemoglobinuria due to cold.

Herrick.

Hypersplenism.

Israel.

Jaffe-Gottfried-Bradley.

Magnus.

Marchiafava-Micheli.

Hereditary methemoglobinemia.

Minkowsky-Chauffard.

Moschowitz.

Hereditary hemolytic ovalocytosis.

Post cardiac surgery.

Radiation.

Rietti-Greppi-Micheli.

Thalassemia.

Tibialis anterior.

Tixier.

Uremic-hemolytic.

Widal-Ravaut.

Hemorrhagic syndromes due to alterations in hemostasis

Purpura due to vascular alterations

Lightning purple.

Non-thrombocytopenic purpuric (see vascular fragility).

Vascular purpura.

Rendu-Osler-Weber.

Schönlein-Henoch.

Purpura due to extravascular abnormalities

Ehlers-Danlos.

Epidermolysis bullosa.

Bochelor or Bachelor's scurvy.

Favre.

Gardener-Diamond.

Groenblad-Strandberg-Touraine.

Hyperviscosity.

Majocchi.

Factual purple.

Pruritic purpura.

Psychogenic purple.

Schamberg.

DNA sensitivity.

Touraine.

Waterhouse-Friederichsen.

Purpura due to intravascular abnormalities

Qualitative platelet abnormalities.

Atrombia.

Bernard-Soulier (platelets with morphological changes).

Platelet deficiencies (thrombocytopenic syndromes).

Glanzman.

Glanzman-Naegely (small platelets with agglutination).

Hegglin.

Hermanski-Pudlak.

Hypersplenism.

Kasabach-Merritt.

Myeloproliferative.

Minot-Von Willebrand.

Moschowits.

Post-transfusion purpura.

Radiations.

Hemorrhagic thrombocytopenia.

Thrombocytosis.

Vaquez-Osler.

Werlhof.

Wiskott-Aldrich.

Coagulation disorders

Christmas.

Factor VII deficiency.

Fibrinogenopenia.

Prothrombin deficiency.

X factor deficiency.

Factor XII deficiency.

Classic hemophilia.

Hemorrhagic of the newborn.

Parahemophilia.

Syndromes with hypersplenism

I Secondary (symptomatic)

Banti syndrome.

Non-Hodgkin's lymphomas.

Hodgkin's disease.

Rheumatoid arthritis, Felty syndrome, collagenosis,

Gaucher disease, Nieman Pick, etc.

Sarcoidosis, amyloidosis.

Histiocytosis

Kala-Azar.

Malaria, tropical splenomegaly.

Chronic infections, tuberculosis, brucellosis.

Thalassemias.

Chronic lymphoid leukemia, myelosclerosis.

Thrombocytopenic purpura.

Infectious mononucleosis.

II Primary (idiopathic)

Non-tropical primary splenomegaly.

Syndromes with myoglobinuria

Bywater.

Haff.

Idiopathic paroxysmal myoglobinuria.

Overuse (overuse syndrome).

Neutropenic syndromes

Treble

Schultz.

Chronics

Congenital agranulocytosis.

Pediatric genetic agranulocytosis.

Felty.

Reticular dysgenesis.

Doan-Wiseman.

Chronic granulocytopenia in children.

Marchiafava-Micheli.

Myelofibrosis.

Cyclic neutropenia.

Familial neutropenia.

Chronic hypoplastic neutropenia.

Neonatal neutropenia.

Schwachman.

Syndromes with polycythemia

Yesterday.

Chronic pulmonary heart.

Congenital heart diseases.

Benign familial erythrocythemia.

Gainsböck.

Hypoventilation.

Monge.

Mosse.

Polycythemia associated with tumors.

Neonatal polycythemia.

Vaguez-Osler.

Thalassemic syndromes

According to the type of globin altered.

Alpha thalassemia.

Beta thalassemia.

Thalassemia-F.

Delta thalassemia.

Lepore thalassemia.

Different combinations of hemoglobin alteration result in the syndromes described above. The combination of thalassemia with other types of hemoglobin causes other syndromes (see syndromes due to hemoglobinopathies).

CAT SCRATCH SYNDROME

Synonymy

Cat scratch fever. Cat scratch disease. Elorudodruphitis. Cat scratch disease.
Benign lymphoreticulosis

Syndromography

Clínic

It is a subacute, circumscribed disease, characterized by discomfort, granulomatous lymphadenitis and variable types of fever, which is usually preceded (between 3 and 14 days) by a cat scratch that produces a pustular lesion (sometimes red, painful papules) that is accompanied, in the following two weeks, with damage to the regional chain corresponding to the lymph nodes.

The inoculation site (red papule) is seen in most cases (50-90%). The occurrence of purpuric-type lesions (thrombocytopenic purpura), encephalitis, and osteolytic lesions has been reported.

Paraclinical examinations

The diagnosis is based exclusively on the clinical picture and the histopathological characteristics of the affected lymph nodes. The pus obtained from the lymphatics does not show bacterial growth.

Peripheral blood. Lymphocytosis with left shift of polymorphonuclear cells.
Immunology. The Foshay antigen skin test has been reported to be positive.

Syndromogenesis and etiology

No etiological agent has been identified, but the presence of pleomorphic gram-negative bacilli that stain with silver has been described in the affected lymph nodes.

Literature

Phillips, W.R.: "Cat-scratch syndrome." New York J. Med., 54:1769,1954.

Benenson, A.: The control of communicable diseases in man. Scientific Publication No. 507 PAHO/WHO, 1987. p.101.

BAR SYNDROME

Synonymy

Colibacillosis gravidarum.

Syndromography

Clinic

It is characterized by abdominal pain often located in the gallbladder, uterine or appendicular region and fever that appears during pregnancy, although sometimes it can occur in the postpartum period.

Paraclinical examinations

Urine cultures and blood cultures. They demonstrate 1the presence of colibacilli.

Syndromogenesis and etiology

This is a condition caused by bacillary septicemia whose starting point is the colon.

GRAM NEGATIVE SHOCK SYNDROME

Synonymy

Endotoxic shock due to gram-negatives.

Syndromography

Clínic

This is a serious condition, which occurs more frequently in the elderly, young children and people affected by diabetes, liver cirrhosis or malignant tumors. It has a sudden onset with fever and chills, alternating periods of alertness and lethargy, tachycardia, tachypnea and hypotension and very rarely nausea, vomiting or diarrhea. On the skin it is possible to observe brownish mottled type lesions; 30 9% progress to shock and the mortality rate can reach up to 80%.

Paraclinical examinations

Blood. Leukocytosis, but leukopenia is observed in Pseudomonas infections. Metabolic acidosis.
Pathological anatomy. Congestion of the vascular bed of the spleen and liver, areas of necrosis and hemorrhages in the intestinal tract.

Syndromogenesis and etiology

The etiology of this syndrome is related to the release of endotoxins from gram-negative germs during bacteremia caused by these agents (E.coli, Proteus, Aerobacter aerogenes, P. aeruginosa, etc.).

Literature

Waisbren, B.A.: "Gram negative shock and endotoxin shock." Amer. J. Med. 36:819-824, 1964.

TOXIC SHOCK SYNDROME

Syndromography

Clinic

It was initially described in 1978 and it was known that it could cause a clinical picture of high fever and similar erythema.

to a sunburn, accompanied by palmoplantar peeling and hypotension. Alterations of at least three different organs or systems must be associated. Vomiting and diarrhea, signs of kidney or liver failure, myalgia, disorientation, etc. are common. In 1980, an epidemic form was described among young women (mainly white) who began menstruation, associated with the use of certain hyperabsorbent sanitary "tampons."

The guidance given to the public and the reduction in the use of hyperabsorbent tampons have decreased cases of this syndrome.

Paraclinical examinations

Blood. Thrombocytopenia. Elevation of creatine phosphokinase.

Cerebrospinal fluid. Normal

Syndromogenesis and etiology

A very close association has been found with the presence of Staphylococcus aureus in vaginal and cervical cultures and later a toxin called TS-CT-1 was identified, which is produced by staphylococci and is thought to be the mediator of this syndrome, although the possibility of the existence of other toxins has not been ruled out.

Literature

Chambers, H.F. et al.: "Staphylococcus aureus endocarditis: Clinical manifestations in addicts and nonaddicts". Medicien, 62:170, 1983.

Garbe, P.L. et al.: "Staphylococcus aureus isolates from patients with nomenstrual toxic shock syndrome. Evidence for additional toxins." J.A.M.A., 253:2538,1985.

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DENNIE-MARFAN SYNDROME

Synonymy

S. of congenital syphilitic paralysis. S. of Marfan.

Syndromography

Clinic

It is characterized by the triad: paralysis (spastic or flaccid), mental retardation and congenital syphilis.

Paraclinical examinations

Examination in dark field or by phase microscopy. Exudates from lesions or aspiration material from lymph nodes show Treponema pallidum.

Serological tests (VDRL). They can be performed in blood and/ or cerebrospinal fluid; the result is positive.

Syndromogenesis and etiology

It is caused by transplacental maternal-fetal (congenital) infection of Treponema pallidum (spirochete that produces syphilis). Congenital transmission of syphilis is more likely in the presence of early maternal syphilis, but it can also occur throughout the latency period.

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TROPICAL SPLENOMEGALY SYNDROME

Synonymy

Idiopathic splenomegaly. Giant spleen. Cryptogenic splenomegaly. Bengal splenomegaly. S. of African macroglobulinemia.

Syndromography

Clinic

The fundamental finding is a marked splenomegaly associated with asthenia and intense fatigue that is observed in inhabitants, predominantly women, of tropical and subtropical regions with malaria endemic.

Paraclinical examinations

Blood. One characteristic is that anemia is always present. Parasitological investigations for malaria are negative or very few parasites are observed.

In Nigeria, lymphocytosis is detected in peripheral blood and bone marrow. In Uganda and New Guinea lymphocytosis is absent.

In all cases there is an increase in serum immunoglobulin (IgM).

Spleen biopsy. The characteristics of the spleen vary in different regions: in New Guinea, greater dilation and less lymphatic proliferation is found than in cases on the African continent. There is absence of malarial pigment.

Syndromogenesis and etiology

Malaria parasites are considered direct or indirect agents but there is no definitive conclusion in this regard. The existence of an abnormal immune response is proposed. There is an acceptable therapeutic response to antimalarial treatment (but this has to be prolonged) even in the absence of the parasites. Cessation of treatment is often followed by a relapse of the syndrome within three months.

The etiology is unknown.

Literature

Conferences and Meeting: Tropical splenomegaly syndrome. Brit. Med. J., 4:614, 1967.

CHRONIC FATIGUE SYNDROME

Synonymy

S. of myalgic encephalomyelitis. Post-viral fatigue syndrome. Chronic Epstein-Barr virus infection.

Syndromography

Clinic

Chronic fatigue syndrome is a relatively common clinical disorder, which mainly affects adults between 20 and 50 years of age.

and it is fundamentally characterized by severe fatigue induced by physical activity, sometimes minimal. After the onset of the symptomatic crisis, the individual generally recovers within hours or days.

Fatigue is associated with the inability to perform tasks that were previously performed easily, such as walking to school or work, shopping, etc. The patient describes his symptoms as a weight on the muscles, but not pain.

Additionally, neurological or psychiatric symptoms may occur, such as difficulty concentrating, depression and anxiety or loss of fixation memory (short time).

Other symptoms that may be found are myalgia, arthralgia, headache and insomnia. Most cases have recently suffered an acute infectious process.

The medical literature records an epidemic behavior of this syndrome, reporting outbreaks that have affected dozens and hundreds of people in numerous countries. One of the best-studied outbreaks occurred in London in 1955 and affected almost 300 people, including doctors, nurses and other workers and patients at the Royal Free Hospital. Currently, the epidemiological presentation predominates in the form of sporadic cases.

The criteria to accept the diagnosis of this syndrome include:

1. Chronic, persistent or recurrent fatigue (exacerbated by light physical exercises that cause the impossibility of performing usual tasks, for a period greater than 6 months).
2. Neuropsychiatric dysfunction (loss of ability to concentrate, inability to make mental efforts, loss of memory of events that occurred in a short time).
3. Exclusion of another alternative diagnosis (clinical history and appropriate investigations).

Syndromogenesis and etiology

The etiology has not been precisely specified, although the presence of infections by numerous agents has been established as probable triggering factors of the clinical picture, among them the following have been noted: enterovirus, influenza virus, varicella virus and Epstein-Bar virus. It has also been suggested that infections caused by toxoplasma and brussels can induce the syndrome.

More recently, herpesvirus 6 (HHV-6) has been reported. The concept that the permanence of viruses or viral remnants can cause chronic fatigue syndrome is supported by the finding of high and persistent serum levels of antibodies to viruses such as Coxsackie and Epstein-Barr. Using modern techniques for detecting RNA, the presence of viral material has also been verified in the muscles of many of these patients.

Literature

Lloyd, A.; D. Wakefield; C. Boughton and J. Dwyer,: "What is myalgic encephalomyelitis? Lancet", 1:1286-1287, 1988.

Holmes, G; J. Kaplan; N. Gantz et al."Chronic fatigue syndrome: a working case definition". Ann. Intern. Med. 108:387-389, 1988.

BULLIS FEVER SYNDROME

Synonymy

Tick fever.

Syndromography

Clinic

It is an acute condition, with a sudden onset with fever and chills and lasting between 4 and 14 days. This is followed by intense occipital headache, malaise, anorexia, and an intense 48-hour maculopapular rash and lymphadenopathy.

Paraclinical examinations

Blood. Leukopenia with neutropenia.

Syndromogenesis and etiology

It is thought to be due to a species of Rikettsia, probably transmitted by tick bites.

PFEIFFER FEVER SYNDROME

Synonymy

Infectious mononucleosis. Kissing disease. S. de Filatou. S. de Turck. Gamma herpesvirus mononucleosis.
Epstein-Barr virus mononucleosis. Ganglion fever.

Syndromography

Clínic

This syndrome is clinically characterized by an acute symptoms of fever, pain and inflammation of the throat (often with exudative pharyngotonsillitis) and lymphosplenomegaly (especially of the posterior cervical chain) in 40-60% of cases. One of these three elements of the triad dominates the clinical picture and the others may be present to a lesser extent or absent.

Other findings are: eyelid edema (12%), conjunctivitis, optic neuritis, keratitis and a rubella-like rash (15%), meningoencephalitis or Guillain-Barré syndrome (1%), jaundice (5%) and rarely hemorrhagic manifestations. It has a worldwide distribution and is very common in young children in underdeveloped countries where it is usually mild or asymptomatic. The typical form is seen more in developed countries where the age at which infection occurs is delayed until it affects older children and young adults.

Paraclinical examinations

Blood. Lymphocytosis greater than 50% (including 10% or more abnormal forms).

Immunofluorescence test. To detect IgM or IgA antibodies against viral Capsid or early onset antibodies against the causal virus.

Hemagglutination test for immune adhesion. It is very sensitive and specific.

Syndromogenesis and etiology

It is produced by the Epstein-Barr virus, a 4 (gamma) herpes virus closely linked in its morphology to other herpes viruses, but with serological differences, which infects and transforms B lymphocytes. It spreads from one person to another through the oropharyngeal route, through saliva. Kissing facilitates transmission between adults. Transmission through blood is also possible.

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Joncas, J.: *Infectious mononucleosis". Cand. Med. Ass. J.. 96:1212- -1216, 1967.

PRETIBIAL FEVER SYNDROME

Synonymy

S. of Fort-Bragg.

Syndromography

Clinic

The occurrence of sporadic cases has been reported in the states of North Carolina and Georgia (USA). It has a sudden onset with general malaise, headache, moderate non-persistent photophobia, respiratory symptoms, chills, and fever in the form of spikes two or more times a day, for 4-8 days.
Around the fourth day of the illness, an erythematous rash appears, limited to the anterior region of both legs, which usually remains for 2 or more days and leaves residual pigmentation that lasts 2 weeks. Splenomegaly and relative bradycardia are also observed.

Paraclinical examinations

Blood. Initial leukopenia followed later by moderate leukocytosis with relative lymphocytosis. Complement fixation for positive leptospira.

Syndromogenesis and etiology

Leptospira autumnalis and Leptospira pomona have also been observed in association with a similar pretibial syndrome, but with generalized rash.

Literature

Daniels, W.B. and H.A. Grennan: “Pretibial fever.” J.A.M.A.. 122:361-365, 1943.
Sanford, J.P.: Leptospirosis. In: Beeson, P.M., and W. McDermott, eds:. Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia, Saunders, 1967.

GREGG SYNDROME

Synonymy

S. of congenital rubella.

Syndromography

Clinic

It is a condition of congenital malformations of the fetus caused by the mother's rubella virus infection during the first trimester of pregnancy. During the physical examination, the following malformations can be observed (alone or associated): microcephaly, microphthalmia, bilateral cataracts (which produce blindness), deafness, cardiac malformations and mental deficit. These malformations can occur even if the maternal rubella case was asymptomatic.

Paraclinical examinations

Blood. Positive VDRL test. Specific IgM antibodies in a single sample.

Viral culture for rubella. Positive pharyngeal samples.

Syndromogenesis and etiology

Rubella virus infection during the first trimester of pregnancy.

Literature

Benenson, A.: The control of communicable diseases in the man. Scientific Publication, No. 507 PAHO/WHO, 1987, p.395.
Lande, L.: "Congenital marformations with severe damage to central nervous system due to early fetal virus infection." J. Pediat..36:625, 1950.

PULMONARY HANTAVIROSIS SYNDROME

Syndromography

Clinic

A specific hantavirus has recently been identified as the cause of an outbreak of respiratory disease (hantavirus pulmonary syndrome [HPS]). The syndrome was initially recognized in the southwestern United States in May 1993. The first retrospectively confirmed cases of HPS occurred in July 1991, and the two most recent in September 1993.

The age of the patients has ranged between 12-69 years with an average of 32 years. 52% were male and 62% of the cases died.

The syndrome is characterized by a prodromal period accompanied by fever, myalgia and variable respiratory symptoms, followed by an abrupt onset in the form of acute respiratory distress. Other symptoms reported during the initial phase include headache and gastrointestinal manifestations (abdominal pain, nausea and vomiting). During its evolution, arterial hypotension and signs of hypoxia appear accompanied by fever.

No sequelae were seen in the survivors.

Paraclinical examinations

Blood. Hemoconcentration (76%), trompocytopenia (71%).

Chest radiology. In all cases there is pulmonary inflammatory infiltrate from the first days of hospitalization.

Pathological findings. Edematous lungs that, upon microscopic examination, show interstitial infiltration of mononuclear cells in the alveolar septa, as well as septal and alveolar edema with or without mononuclear exudate. Occasionally alveolar hemorrhage. Pleural effusions. In the white pulp of the spleen, portal spaces of the liver and in other viscera, large mononuclear cells with the appearance of immunoblasts are observed.

Histochemical study. Monoclonal and polyclonal antibodies make it possible to detect the Hantavirus antigen in tissues, with marked accumulation in the lungs.

Syndromogenesis and etiology

The cause appears to be Hantavirus, which has been confirmed by the demonstration of specific antibodies in 39 of 42 patient sera samples, particularly to Hill virus.
In 37 cases, the polymerase chain reaction (PCR) revealed Hantavirus RNA in frozen lung tissue.

The reservoir of this virus is represented by a rodent called Peromyscus maniculatus, which is widespread throughout most of the United States, except in the southeastern region.

The clinical syndromes associated with Hantavirus infections, previously known, are characterized by hemorrhagic symptoms with kidney lesions (Bolivian and Argentine hemorrhagic fevers, Junin, Machupo or Arenavirus fever).

In the syndrome caused by Hantavirus, respiratory symptoms predominate and kidney damage is present only rarely. However, the differential diagnosis between HPS and other forms of non-cardiogenic pulmonary edema or adult respiratory distress syndrome is very difficult, especially at the beginning of the clinical picture.

Bibliography

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CDC. "Outbreak of acute illness-southwestern United States." MNWR. v. 42:421-424.
CDC. "Ubdate: outbreak of Hantavirus infection-southwestern" United States. MNWR, 42: pp. 770-771, 1993.

INGUINAL SYNDROME

Synonymy

Inguinal lymphogranuloma. Estiomeno. Climatic bubo. Tropical bubo.

Syndromography

Clinic

This is the most common variant of the clinical picture of lymphogranuloma venereum observed in homosexuals and is characterized by painful inguinal lymphadenopathy that begins between 2-6 weeks after the suspected infective contact and rarely appears a few months later. This adenopathy is unilateral in two out of three cases and on the same side an enlargement of the iliac and femoral nodes can be palpated. Initially small nodes may progress to the formation of a fluctuating, suppurating mass and the underlying skin becomes inflamed, thin, and fistulized. The sulcus sign is frequently observed, which consists of an enlargement of the lymph node chains located above and below the crural arch. This condition may or may not be accompanied by signs of proctitis (anorectal pain, mucopurulent and bloody rectal discharge, tenesmus, etc.).

Paraclinical examinations

Crops. Positive to Chlamydia trachomatis.

Serology for Ch. trachomatis. Positive. Positive microimmunofluorescence test.

Histology. Infected lymph nodes initially show small, stellate abscesses, surrounded by histiocytes. Demonstration of inclusion bodies in the leukocytes of the fluid aspirated from the bubo.

Syndromogenesis and etiology

The syndrome is produced through sexual contact, by infection with Chlamydia trachomatis (a microorganism that is related to the agent of trachoma and oculogenital infections due to Chlamydia) but with immunotypes L-1, L2 and L-3.

Literature

Benenson, A.: The control of communicable diseases in man. PAHO/WHO Scientific Publication No.507, p. 288, 1987.
Alexander, E.R.; E.R. Harrison: "Role of Chlamydia trachomatis in perinatal infection." Rev. Infect. Dis., 5:713, 1983.
Jones, R.B. et al.: "Clamydia trachomatis in the pharynx and rec tum of heterosexual patients at risk for genital infection." Ann. Intern. Med., 102:757, 1985.

KATAYAMA SYNDROME

Synonymy

Katayama fever. S. of cardio-pulmonary schistosomiasis. S. of the invasive phase of bilarzia. S. cor pulmonale Bilharzian.

Syndromography

Clinic

The most common symptoms that characterize this syndrome are: headache, chills, fever (lasting 1-4 weeks), diarrhea, splenomegaly, hepatomegaly. In addition, there is angioneurotic edema, urticaria, abdominal pain, melena, and sometimes massive hematemesis.

Migration to the lung is manifested by dyspnea. The clinical picture is produced by pulmonary infection caused by Schistosoma mansoni or S. haematobium and is characterized by hypertrophy of the right heart with functional failure that does not respond to the usual treatments, since it is due to pulmonary arteriolitis obliterans, which is produced by the accumulation of parasite eggs in the lumen of the arterioles.

Migration to the brain causes: aphasia, paraplegia, Jacksonian epilepsy, coma and other neurological manifestations.

Physical examination. At the beginning: hepatomegaly, pain on palpation of the liver edge, splenomegaly. Later: liver atrophy, cirrhosis, medusa head sign, ascites, progressive weight loss.

Paraclinical examinations

Biopsy. Eggs in different fabrics.

Stool. Presence of eggs.

Serology, ELISA test, radioimmunoassay, fluorescent antibodies. Antigens purified from eggs or the adult worm are used. It should be clarified that the positive results of these tests do not determine that there is a current infection, since the patient may have suffered from it before and remain positive.

Blood. Marked eosinophilia, anemia, leukopenia, thrombopenia. In short, pancytopenia. Hyprothrombinemia.

Syndromogenesis and etiology

The symptoms are caused by infection produced by various varieties of trematodes. This syndrome is endemic in Africa and the Arabian Peninsula; in Brazil, Suriname and Venezuela, in America, as well as in some Caribbean islands (S. mansoni). The S. haematobium variety is located in Africa and the Middle East and S. japonicum in China, Japan and Southeast Asia.

The symptomatic picture depends on the number and location of the parasites (Schistosoma mansoni. S. haematobium and S. japonicun) in the human host, in this case in the lumen of the pulmonary arterioles. The deposited eggs produce tiny granulomas and scars that increase pulmonary hypertension, which causes the clinical picture.

The infection is acquired through skin contact with water when bathing or wading in different bodies of water (lagoons, rivers, ponds, etc.) that contain larvae (cercariae). The persistence of the parasite in these bodies of water depends on the presence of a snail that acts as an intermediate host (Biamphalaria, Bulinos and Onco-melania).

Literature

Benenson A.: The control of communicable diseases in man. Scientific Publication, No. 507 PAHO/WHO p. 166. 1987.
Walt, F.: "The Katayama syndrome." South Africa Med. J., 28:89-93, 1954.

KAWASAKI SYNDROME

Synonymy

Kawasaki disease. S. of the mucocutaneous lymph nodes. Acute febrile lymphonodular mucocutaneous S.

Syndromography

Clínic

It is an acute febrile condition that occurs in childhood (infants and children under 5 years of age) characterized by peaks of high fever that does not respond to antibiotics and generally lasts 5 days, associated with irritability, non-suppurating cervical adenopathy, bilateral congestion of the conjunctivae, enanthem and rash. The enanthem consists of an eruption of the tongue (strawberry-like tongue) and the exanthema is erythematous, polymorphous and usually affects the trunk.

Edema may also be seen in the limbs. In 15-30% of cases, coronary artery aneurysms occur due to arteritis. The case fatality rate is 1-2%.

Paraclinical examinations

There is no specific test for diagnosing the syndrome.

Blood. Increased erythrocyte sedimentation rate during the acute phase. The number of platelets increases to more than 500,000/mm² during the second and third weeks of the disease and remains elevated for about 3 weeks.

Syndromogenesis and etiology

A history of viral respiratory diseases and contact with mites that are related to some bacterial species have been suggested as possible causal factors.
Epidemic outbreaks of the disease have been reported in US cities with greater frequency in winter and early

Mavera.

The etiology is unknown.

Literature

Benenson, A: The control of communicable diseases in man. Scientific Publication, No. 507 PAHO/WHO p. 425, 1987.

KURU SYNDROME

Synonymy

S. of the smiling death.

Syndromography

Clinic

This condition has been described exclusively in the Fore linguistic group, in highland regions of Papua New Guinea and predominantly affects children over 4 years of age and adult women, to a lesser extent. The symptoms are given by cerebellar ataxia and tremors of the leg muscles, uncoordinated movements of the arms when extending, exaggeration of voluntary movements, shaking, difficulties in articulating words, fecal incontinence and in the end aphonia and dysphagia. The patients, however, have a smiling face,

This condition, which was very common in the past, is currently reported in less than 10 cases per year.

Paraclinical examinations

Blood. Elevated globulin levels.

Virological and immunological studies. It has not been possible to identify a responsible virus, although there are some immunological alterations.

Syndromogenesis and etiology

When patients die, widespread changes are observed in central nervous structures that include demyelination, loss of neurons, proliferation of microglia, and perivascular infiltration.

The syndrome is caused by a filterable agent with the capacity for self-replication, transmittable to primates and other animals.
Some experts have hypothesized that infectious agents are proteins of a peculiar type that have the property of replicating by an unknown mechanism. It is currently classified together with Creutzfeldt-Jakob syndrome as a slow virus infection of the central nervous system. The suggested transmission mechanism is intimate contact with mortuary tissues, especially cannibalism.

Literature

Benenson, A.: The control of communicable diseases in man. OPS. Scientific Publication No 538, p 121,1992.
Gajdusek, D.C. and V. Zigas: Degenerative disease of the central nervous system in New Guinea. The endemic occurrence of Kuru in the native population'". New Engl. J. Med., 257:974-978, 1957.

Gajdusek, D.C.; C.J. Gibbs and M. Alpes: "Experimental transmission of a Kuru-like syndrome to chimpanzees." Nature, 209:794-796, 1966.

LYME SYNDROME

Synonymy

Chronic erythema migrans with polyarthritis. Lyme arthritis.
Tick meningopolyneuritis. Lyme disease.

Syndromography

Clinic

It is characterized by skin lesions, general symptoms, polyarthritis and neurological and cardiac conditions. The disease is more common in the summer and its first manifestation is skin lesions, which consist of red macules or papules that extend in an annular shape, sometimes with multiple similar lesions. Later they are accompanied by discomfort

general, fatigue, headache, fever, neck stiffness, myalgia, migratory arthralgias and lymphadenopathy lasting several weeks.
Weeks or months later, a neurological condition may appear (aseptic meningitis, encephalitis, chorea, cerebellar ataxia, facial paralysis, motor or sensory radiculoneuritis, and myelitis).
Cardiovascular abnormalities such as atrioventricular block, acute myopericarditis or cardiomegaly also occur;
late swelling and pain in the large joints, especially the knees, reappearing for years. It can end up like a certain type of chronic arthritis.

Paraclinical examinations

Skin biopsies.

Blood and cerebrospinal fluid. Identification of the spirochete in lesions.

Titer test for specific antibodies against the causative spirochete. The diagnosis is commonly confirmed with this test.

Syndromogenesis and etiology

It is a transmissible zoonotic disease caused by a spirochete identified in 1982 and called Borelia burgdoferi, which is transmitted by ticks (Lxodes dammini and Lpacificus, in the USA and I.ricinus in Europe). The cycle of this agent is perpetuated in nature through the transmission from one population to another of some species of ticks, deer, wild rodents and other animals.

Literature

Benenson A.: The control of communicable diseases in man. Scientific Publication, No. 507 PAHO/WHO, pp. 99-101, 1987.
Pachner, A.R.; B.C. Steere: "The triad of neurological manifestations of Lyme disease. Meningitis, cranial neuritis, and radiculoneuritis." Neurology, 35: 47, 1985.
Steere, A.C.: S.E. Malawista: Lyme disease, in Principles and Practice of Infections Diseases, ed.2 G.L. Mandell et al. (eds.). New York, Wiley, pp. 1343-1349, 1985.
Johnston, Y. et al.: "Lyme arthritis: Spirochetes found in synovial microangiopathic lesions." Am. J. Pathol., 118:26, 1985.

HAND-FOOT-MOUTH SYNDROME

Synonymy

S. of late-year blisters.

Syndromography

Clínica

It mainly affects farmers and people who are in contact with livestock and is characterized by a vesicular-type rash restricted to the hands and feet, and an oral enanthema that affects the uvula, pillars and tongue. It is accompanied by fever, general malaise, anorexia, diarrhea, and sore throat. Occasionally there is hepatosplenomegaly and lymphadenopathy.
Small ulcerative lesions can be found in the mouth, which occur when the vesicles rupture. The evolution is towards recovery.

Paraclinical examinations

Viral isolation and identification. In samples from the throat and rectum, or blood.

Paired sera. During illness and convalescence.

Syndromogenesis and etiology

The syndrome is the result of infection with coxsacke virus group A type 16 and other groups.

Literature

Cherry, J.D. and CL. John: "Hand, foot, and mouth syndrome: report of six cases due to coxsackie virus. Group A type 16." Pediatrics, 37:637-643, 1966.
Robinson, R.; EW. Doane and A.J. Rhodes: "Report of an outbreak of febrile illness with laryngeal lesions and exanthem: Toronto, summer 1957; isolation of group A cOxsackie virus." Canada Med. Ass. J., 79:615-62 1, 1958.

MENINGOCOCEMIA SYNDROME WITHOUT MENINGITIS

Synonymy

S. of chronic meningococcemia.

Syndromography

Clínic

It occurs in relatively healthy individuals, with fever, malaise, pain in the joints, followed, a few days later, by rash, erythematous macules in different parts of the body (frequently on the palms of the hands and soles of the feet), in which irregular hemorrhagic areas develop whose most characteristic forms are pink lesions with petechiae in the center. It is accompanied by splenomegaly.

Paraclinical examinations

Blood culture. Meningococci.

Skin biopsy (36-48 hours). A perivascular infiltrate of lymphocytes and macrophages, few granulocytes and absence of fibrin or occlusive thrombi is observed. The vascular walls are intact and no endothelial thickening or edema is observed in the dermis; the epithelium is normal. Unlike the acute form of meningococcemia, there is an absence of bacteria.

Syndromogenesis and etiology

The condition is caused by meningococcus and it has been suggested that it occurs in individuals with partial immunity to this agent.

Literature

Salomon, H.: Ueber Meningokokenseptikamie'". Klin. WSchr.. 39:1045-1048, 1902.
Ognibene, A.J. and W.R. Dito: Chronic menigococcemia". Arch. Int. Med., 114:29-32, 1964.

MYCOBACTERIA-PANCITOPENIA SYNDROME

Syndromography

Clínica

It presents as a prolonged clinical condition with periods of relapses and remissions, characterized by fever, weight loss, sweating, anorexia and lung infections.

Physical examination. The main findings are: sweating, hepatomegaly and/or splenomegaly and adenomegaly.

Paraclinical examinations

Blood. Severe anemia, thrombocytopenia, leukopenia with neutropenia (sometimes leukocytosis may be present).

Medulogram. Generally spinal hypoplasia or increased erythropoiesis.

Pith culture. atypical mycobacteria

Tuberculin test (PPD). Negative or weak-positive.

Pathological anatomy. Miliary type pulmonary infiltrate in 30% of cases, other lung infections are also common. Osteolysis and granulomatous lesions in lymph nodes, pancreas, spleen, liver and kidneys.

Syndromogenesis and etiology

The etiology of this condition is related to the infection of unidentified mycobacteria, coinciding with a myoloproliferative process that resembles lymphomas.

Literature

Kilbridge, TM.; J.S. Gonnella and J.T. Bolan: "Pancytopenia and death." Arch. Int. Med., 120:38-46, 1967.

MCLEOD SYNDROME

Syndromography

Clinic

It is caused by the appearance of signs and symptoms of pneumonmediastinum and subcutaneous emphysema and represents a late complication in the course of measles pneumonitis.

Paraclinical examinations

Chest radiology. A unilateral radiolucent area of the lung is observed.

Syndromogenesis and etiology

The cause is the measles virus.

Literature

Hoeprich, Paul de: Treatise on infectious diseases.t. 1, Revolutionary Editions p.699, 1982.

GREEN URINE SYNDROME

Synonymy

S. of toxemia due to Pseudomonas.

Syndromography

Clínic

It is a very serious condition, often fatal, that is observed in patients who present extensive lesions that become infected with P. aeruginosa (empyemas, large burns, etc.) and is characterized by the appearance of green urine preceded a day or two by hemolytic crisis, hypothermia, oliguria and dehydration with or without shock.

Paraclinical examinations

Urine. Presence of green bile pigments (greenhemnoglobin) must be differentiated from other pigments that Pseudomonas infection also produces.

Pus culture. Pseudomona aeruginosa is identified.

Blood. Decreased hematocrit. Hyperbilirubinemia direct.

Syndromogenesis and etiology

Massive infection by P. aeruginosa causes massive hemolysis and formation of greenhemoglobin.

Literature

Stone, H.H.: "The green urine syndrome: an ominous manifestation of pseudomonas toxemia." Bull. Emory Univ. Clin., 3:81-86, 1964.

PAROTIDITIS-HERPANGINA SYNDROME

Synonymy

coxsackievirus S. Lymph nodular pharyngitis. Enteroviral lymphonodular pharyngitis. Vesicular stomatitis with rash.

Vesicular pharyngitis.

Syndromnography

Clínic

It is a self-limiting clinical condition, characterized by inflammation of the parotid glands and ulcerative or vesicular lesions of the fauces associated with coxsackie virus infection. Pharyngeal lesions are circumscribed (1-2 cm), papulovesicular and grayish on an erythematous base and may eventually ulcerate.

Paraclinical examinations

Virology. Viral isolation in lesions and samples of nasopharyngeal material and feces of lactating mice.

Syndromogenesis and etiology

Coxsackievirus, group A, types I to 6, 8, 10 and 22. Other enteroviruses have occasionally been related to this clinical picture.

Literature

Benenson, A.: The control of communicable diseases in man. Scientific Publication, No. 507 PAHO/WHO, p.107, 1987.

REITER SYNDROME

Synonymy

S. de Fiessinger-Leroy-Reiter. S. of urethritis arthritis-conjunctivitis. S. de

Syndromography

Clinic

This syndrome has been described in the form of a triad: arthritis, conjunctivitis, urethritis.

It predominates in men and begins between the second and fourth ta decades. It is characterized by: urethritis, with few symptoms or more rarely intense pain during urination, hematuria and purulent discharge; This is frequently the first manifestation of the symptomatic triad. Circinate balanitis later develops. Conjunctivitis in 50% of patients, occurring about 10 days after urethritis. Its intensity varies from

a simple congestion to purulent discharge. It is rarely accompanied by iritis or keratitis. Arthritis is the most constant element of the triad. It is acute, asymmetric and oligoarticular, and primarily affects joints of the lower limbs, especially knees, ankles, metatarsals and interphalanges of the toes. It is accompanied by malaise and fever. The joints are hot, erythematous and painful. Inflammation of the plantar aponeurosis and Achilles tendon are common. The crisis lasts from days to months and relapses triggered by sexual contact followed by urethritis are common.

Permanent injuries may remain, such as: heel spurs, cavus or flat feet, dorsiflexion, etc.

Mucocutaneous lesions are common and appear in the mouth, glans, palms of the hands, and soles of the feet.

Superficial lesions of the mucosa of the mouth and glans are painless and are observed in 33% of cases.

Even in 30% of individuals with the postvenereal form, there is gonorrhagic keratoderma, which consists of crusted, ruptured papules and vesicles on the palms, soles, and glans; To a lesser extent they can be observed on the trunk and limbs, scalp and scrotum; There is cornified material in the nails that raises the nail plate, but there are no "dimples" in the nail plate, which differentiates it from psoriatic arthritis.
The scabs and horny masses of the skin tend to peel off from the surface and leave hyperpigmented spots.

Two clinical forms have been proposed: postvenereal and postdysenteric or epidemic (reactive arthritis). Both clinical forms usually begin with urethritis.

Physical examination. Erythema, pain and heat. There are mucocutaneous lesions in the mouth, glans, palms of the hands and soles of the feet. More rarely, signs of pleuropericarditis, neurological manifestations and secondary amyloidosis may appear.

Paraclinical examinations

Blood. Mild anemia, leukocytosis, accelerated erythrocyte sedimentation rate, increased C-reactive protein.

Synovial fluid. It is inflammatory, with a predominance of polymorphonuclear cells and high complement.

Immunological study. Negative rheumatoid factor. There are no antinuclear antibodies. Determination of HLA B27.

Erythrosedimentation. Elevated in the acute phase.

Radiology. Early cortical erosion, bone neoformation and joint destruction followed by osteoporosis.

In chronic cases there are bone erosions. Apposition of periosteal bone along the diaphysis.

Skin biopsy. Lesions similar to pustular psoriasis, hyperkeratosis.

Syndromogenesis and etiology

Two clinical forms of Reiter syndrome have been identified:

the postvenereal form and the postdysenteric or epidemic form, also known as "reactive arthritis."

The former prevails in North America and Europe, while the post-dysenteric form is more common in developing countries. It is the most common cause of arthritis in young men and an important correlation is found with the presence of the HLA-B27 antigen in them. Even 90% of individuals with Reiter syndrome show positivity for the HLA-B27 antigen.

It is considered that a urogenital or intestinal infectious process, together with a specific genetic predisposition in some people, could trigger the condition.

The most common agents are: gonococcus, Chlamydia trachomatis, Mycoplasma. Shigella dysenteriae and S.flexneri in the intestinal form.

It is considered an autoimmune etiopathogenesis.

Literature

Fiessinger, N. and E. Leroy: "Contribution á l'étude d une épidemie de dysenterie dans la Somme (Juillet-Octobre 1916)".
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Martin, D.H. et al.: "Clamydia trachomatis infections in men with Reiter's syndrome"
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Reiter, H.: Ueber eine bisher unerkannte Spirochateninfektion (Spirochactosis arthritica)". Deutsch. Med. Wschr., 42:1535- -1536, 1916.

Weinberger, H.J.: "Reiter's syndrome." Postgrad. Med., 45:160- -167, 1969.

WATERHOUSE-FRIEDERICHSEN SYNDROME

Synonymy

S. de Marchand-Waterhouse-Friederichsen. S. adrenal meningococcal. Fulminant purpuric meningococcus.

Syndromography

Clinic

It is more common in children, although it can also be seen in adults. It is characterized by irritability, headache, nausea, vomiting, abdominal pain and diarrhea. The fever is moderate at first, but can later be high.

Physical examination. Among the most frequently observed signs are paleness, slimy or sticky skin, disseminated petechiae and ecchymoses, signs of dehydration, dyspnea, tachycardia, hypotension, oliguria; Later, neck stiffness, convulsions, coma, and death occur.

Paraclinical examinations

Crops. Positive to Neisseria meningitidis.

Ionogram. Low chlorides and sodium, high potassium.

Syndromogenesis and etiology

In the vast majority of cases this syndrome is caused by acute infection of the adrenal glands by Neisseria meningitidis, although it has also been observed in other systemic infections such as diphtheria, pneumococcus, staphylococcus and smallpox.

Literature

Frawley, T.EL: In: Eisentein, A.B., ed.: The Adrenal Cortex.
Boston, Little, Brown, 1967.

Waterhouse, R.: "A case of adrenal apoplexy." Lancet, 1:577-578, 1911.

WEIL SYNDROME

Synonymy

S. de Fielder II. S. de Landouzy, S. de Mathieu. S. de Vasilev. Dog fever. Spirochetal jaundice. Silt fever.
Porcine leptospirosis. Weil's disease. Icterus-hemorrhagic leptospirosis.

Syndromography

Clinic

Symptoms include fever, headache, chills, severe discomfort, vomiting, myalgia, conjunctival injection. In

Occasionally, jaundice, renal failure, hemolytic anemia, and bleeding of the skin and mucous membranes are observed. Meningitis and uveitis may also occur.

The duration of these symptoms varies between a few days and three weeks. It mainly affects males, adolescents, young people and middle-aged adults. It is more common during the hot months and mainly in people directly or indirectly exposed to secretions from a wide variety of domestic or wild animals, and in certain countries it has a marked occupational orientation.

Paraclinical examinations

Dark field microscopy. Leptospires are observed.

Blood. Variable from leukopenia to marked leukocytosis with neutrophilia of 70% and anemia. Sometimes thrombocytopenia, hyperbilirubinemia and azotemia. Hypercoagulability is common.

Bacteriology. Serological studies and cultures for leptospires are positive.

Urine. Hematuria, proteinuria and urobilinuria.

Cerebrospinal fluid. In cases that present jaundice, the fluid is xanthochromic, which contrasts with other types of jaundice, which is why it has great diagnostic value.

Syndromogenesis and etiology

Generally, this syndrome is caused by Leptospira icterohemorrhagiae, but it can also be caused by other leptospires (generally, with less severe symptoms), of which more than 170 varieties belonging to about 20 serogroups have been identified.

Literature

Benenson, A.: The control of communicable diseases in man. Scientific Publication, No. 507 PAHO/WHO p. 285, 1987.

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Sanford, J.P.: Leptospirosis. In Beeson, P.B., W. McDermott, ed.: Cecil-Loeb Textbook of Medicine. ed. 12, Philadelphia, Saunders, 1967.

AGAMMAGLOBULINEMIA SYNDROME

ACQUIRED

Syndromography

Clinic

It affects both sexes, begins in adults. There is susceptibility to sinusitis and recurrent pneumonia, chronic progressive bronchiectasis. Spuriform syndromes, proteinorrheic enteropathy.
Arthritic manifestations (rare), exfoliative dermatitis, nephrotic syndrome (occasionally).

Physical examination. Hepatosplenomegaly (common), evidence of non-calcified granulomas in the lungs, skin, liver, spleen (without being able to consider a particular microorganism).

Syndromogenesis and etiology

It is associated with lymphatic malignancies or other dysglobulinemias. Its appearance in relatives suggests that there may be a hereditary problem. (See secondary immune deficiency syndromes.)

Literature

Rosen, F.S. and C.A. Janeway: "Diagnosis and treatment of antibody deficiency syndromes." Postgrad. Med., 43:188-194, 1968.

CONGENTTA AGAMMAGLOBULINEMIA SYNDROME

Synonymy

S. of antibody deficiency.
(See immune deficiency syndromes.)

Syndromography

Clinic

It occurs around the ninth month of life. There is increased susceptibility to pyogenic infections; manifestations of atopy. Difficulty progressing due to frequent infections. If they survive, they develop chronic bronchiectasis, rheumatoid-like arthritis (even preceding infections), and other conditions similar to other collagen disorders.

Syndromogenesis and etiology

In classic agammaglobulinemia, pre-B cells are present in the bone marrow in normal numbers, but their ability to mature appears to be blocked. In children who suffer from the defect, the clinical presentation with recurrent infections is delayed until the second or third year of age.

It presents X-linked inheritance with the gene located at Xq21.3-q22.

Literature

Malcolm, S. et al.: "Linkage of Bruton's aganmaglobulinemia (Imd1l) to probes in Xq21.3-q22: evidence for a second gene coding X for X linked aganmaglobulinemia." (Abstract) Cytogenet. Cell. Genet. 51:1038, 1989.

SWISS TYPE AGAMMAGLOBULINEMIA SYNDROME

Synonymy

S. of thymic dysplasia.

Syndromography

Clínica

Both sexes are affected equally from birth.

Development is delayed and patients present with repeated infections, as well as chronic diarrhea, oral candidiasis, pneumonia, and chronic otitis media.

Infants are susceptible to infections with Candida, cytomegalovirus, and Pneumocystis carini.

Paraclinical examinations

Blood. Lymphopenia less than 2,000, anemia.

Immunological study. Decrease in IgG, IgM and IgA. No response to delayed hypersensitivity tests.
No graft rejection. All T cell immunity tests are abnormal.

Syndromogenesis and etiology

This syndrome is transmitted by autosomal recessive inheritance.

An inability to differentiate cells has been suggested. the progenitors of the marrow, in T and B cells.

The etiology is unknown.

Literature

Amman, A.: "Immunodeficiency diseases." Basic and clinical immunology. 5th. ed., 1985.
Baradun, S.: Das Antikorpermangelsyndrome. Basel, Benno, Schwabe, 1959.
Sell, S.: "Immunological deficiency diseases." Arch. Path., 86:95- -107, 1968.

TEMPORAL ARTERITIS SYNDROME

Synonymy

S. of cranial arteritis. S. of giant cell arteritis. S. of occult temporal arteritis. S. of old man's arteritis. S. of Horton- -Magath.

Syndromography

Clinic

It occurs in elderly people in the seventh or eighth decades of life, rarely before age 55, in both sexes.

At first there is anorexia, insomnia, weight loss, night sweats, light or moderate fever. Temporary pain in muscles and joints.

Classically, fever, anemia, accelerated erythrocyte sedimentation rate, asthenia and headaches in the elderly have been described.

It is related to polymyalgia rheumatica syndrome (pain in the muscles of the neck, shoulders, lower back, hips and thighs).

Injury to the temporal artery is characterized by headache.

At the beginning of the disease there is pain on palpation and pulsations in the artery, which later becomes occluded.

Other times pain is detected in the scalp with claudication of muscles of the lower jaw and tongue. A complication is eye injury secondary to ischemic optic neuritis with sudden blindness in some cases.

The following have been reported: claudication of limbs, cerebrovascular accidents, heart attacks and aneurysms.

Physical examination. Painful increase in volume in the temporal artery area; arteries thick, prominent, often without pulses.

You can have a spontaneous remission between 4-24 months.

Treatment with adrenal steroids manages to alleviate symptoms, anemia and erythrocyte sedimentation rate.

Paraclinical examinations

Blood. very accelerated erythrocyte sedimentation rate, above 100; leukocytosis with left shift in the leukocyte formula; hypochromic anemia that does not respond to treatment. Increase in alkaline phosphatase.

Immunological study. Increased IgG. Excess circulating immunocomplexes. Increased alpha 2 globulin.

Artery biopsy. Thickening of the intima with marked or total reduction of the lumen of the artery or some of its branches. Infiltrated in round cells, eosinophils that simulate tuberculoid granulomas with giant cells (Gilmour).

Syndromogenesis and etiology

It is an inflammation of the medium or large caliber arteries, with injury to one or more branches of the carotid, particularly the temporal one. However, it is a systemic disease and affects multiple sites, immunopathogenic mechanisms (cell-mediated immunity) intervene in it.

It is generally part of a diffuse arteritis: granulomatous arteritis, "giant" cell arteritis.

The etiology is unknown. Currently, it tends to be considered, like polymyalgia rheumaica with which it often coexists, as a collagen disease.

Literature

Cullen, J.F: “Occult temporal arteritis.” Brit. J. Ophthal., 51:513-525, 1967.

Horton, B.T.; TB. Magath and GE. Brown: “An underwritten form of arteritis of temporal vessels." Proc. Mayo Clin., 7:700, 1932.

Hutchinson, J.: "Diseases of arteries." Arch. Surg. (London), 1:323, 1890.

ATAXIA-TELANGIECTASIA SYNDROME

Syndromography

Clínica

It is characterized by a clinical picture of ataxia, oculocutaneous telangiectasias and immunodeficiency. Ataxia begins in lactation and is progressive. There are paranasal sinus and pulmonary infections, the latter recurrent and chronic leading to bronchiectasis.

Patients die from lung diseases or cancers, mainly lymphomas.

Paraclinical examinations

Blood. There is cutaneous anergy.

Immunological study. IgE and IgA deficiency. In few cases there is IgG deficiency (IgG2-IgG4).

LalgMe IgD are within normal levels.

There is an increase in alpha-fetoprotein and carcinoembryonic antigen. normal lymphocytes.

Syndromogenesis and etiology

A defective development of the thymus is proposed.

Patients' lymphocytes respond poorly to T cell mitogens in vitro.

Ataxia can be associated with a generalized defect in cellular differentiation with defects in DNA repair mechanisms.

This syndrome is included with Bloom syndrome and Fanconi pancytopenia within the syndromes with chromosomal instability.

It has an autosomal recessive inheritance.

Literature

Gatti, R.A., et al.: "Ataxia-telangiectasia: an interdisciplinary approach to pathogenesis." Medicine, 70:99-1 17, 1991.

BEHCET SYNDROME

Syndromography

Clínica

The clinical picture is characterized by recurrent, painful, superficial or deep aphthous ulcers with a central necrotic base, isolated or in groups, distributed on the lips, gums, vestibule of the mouth, tongue, tonsils and larynx. They last 1-2 weeks.

They don't leave scars. Ulcerations also appear on the genitals.

It almost always affects young adults.

There is folliculitis, erythema nodosum, acneiform exanthema.

Uveitis that can lead to blindness, iritis, retinal vessel occlusion and optic neuritis. There is also arthritis that affects the knees and ankles and is not deforming.

There may be thrombosis of peripheral veins, benign intracranial hypertension.

Psychiatric disturbances are described.

Paraclinical examinations

Blood. Leukocytosis, accelerated erythrocyte sedimentation rate, elevated Creactive protein.

Immunological study. Increase in circulating immune complexes.

Syndromogenesis and etiology

Bacteria and viruses have been suggested as causal agents, which has not been confirmed. An autoimmune pathogenesis is assumed. Vasculitis exists in almost all patients.

Autoantibodies against the oral mucosa have been identified and circulating immune complexes have been verified.

Literature

O'Duffy, J.O.: "Behcet's disease: Report of 100 cases, 3 with new manifestations." Ann. Inter. Med., 75: 561, 1971.

HEAVY CHAIN SYNDROME

Synonymy

Heavy chain disease. S. of Franklin.

Syndromography

Clinic

The clinical manifestations of this syndrome depend on the isotype of the heavy chain and in relation to this three varieties have been described, which are listed below:

Gamma heavy chain disease or gamma heavy chain disease Franklin

Its clinical picture is similar to that of lymphoma and is characterized by fever, lymphadenopathy, anemia, hepatosplenomegaly and asthenia. Edema of the palate that can [illegible] breathing disorders.

Alpha heavy chain disease or Seligmann's disease

It is the most common and is closely related to Mediterranean lymphoma, a cancer that affects young people in Asia and the Mediterranean.

It is characterized by chronic diarrhea, weight loss, malabsorption, mesenteric and para-aortic lymphadenopathy.

M heavy chain disease

M heavy chain secretion affects a subgroup of patients with chronic lymphocytic leukemia.

Paraclinical examinations

Gamma Heavy Chains

Blood. Demonstration of a component in serum (with levels less than 2 g/100 mL), which reacts in immunoelectrophoresis with anti-IgG, but not with anti-light chains. There may be thrombocytopenia, eosinophilia.

Alpha Heavy Chains

Intestinal biopsy. The lamina propria appears infiltrated by lymphoplasmacytoid cells that secrete alpha heavy chains.

The M component is rare and appears flattened on serum electrophoresis. Immunoelectrophoresis does not detect light chains in serum and urine.

Heavy chains M

Blood. It requires ultracentrifugation or gel filtration and subsequent verification by immunoelectrophoresis of the lack of reactivity of the immunoserum against light chains of said isolated paraprotein.

Syndromogenesis and etiology

They are lymphoplasmacytic cancers. They secrete a defective chain that has an intact Fc fragment and deletion of the Fd region.

There are structural genetic defects with a defect in the assembly of light and heavy chains of the secreted paraprotein.

Literature

Ellman, L.L. and K.J. Bloch: "Heavy chain disease." N. Engl. J. Med. 278:1195-1201, 1968.

Frangione, B. and E.C. Franklin: "Heavy-chain disease: "clinical features and molecular significance of the disordered immunoglobulin structure". Semin. Hematol., 10: 53, 1973.

Franklin, E.C.: J. Lowenstein: B. Bigelow, et al,: "Heavy chain disease of serum gamma globulins. Report of the first case." Amer. J. Med., 37:332-350, 1964.

Osserman, E.F. and K. Takatsuki: "Clinical and immunochemical studies of four cases of heavy (Hgamma 2) chain disease." Amer. J. Med., 37:351-373, 1964.

Selegmann, M.: "Alpha chain disease. Immunoglobulins abnormalies, pathogenesis and current concepts." Br. J. Cancer, 31: 356, 1975.

CHRONIC MUCOCUTANEOUS CANDIDIASIS SYNDROME

Syndromography

Clinic

Unlike candidiasis that occurs in severe deficiencies of cellular immunodeficiency, in mucocutaneous candidiasis syndrome the lesion is superficial and the only main manifestation. Rarely do they develop general infection with Candida or other types of mycoses. Patients are not susceptible to viral or bacterial infections.

Paraclinical examinations

Immunological study. Some patients show immunological defects in the formation of antibodies.

Normal humoral immunity including the ability to make specific anti-Candida antibodies.

Skin tests. There is cutaneous anergy to some variety of antigens and in others only to Candida.

Syndromogenesis and etiology

A group of severely affected patients may be non-genetic. The form of candidiasis with endocrinopathy is a different syndrome. Late-onset cases are not genetic.

Iron deficiency has been suggested, there is a myeloperoxidase deficiency in which there is an increased susceptibility to Candida.

Anergy has been related to the inability of lymphocytes to produce migration inhibitory factor.

Literature

Wells, R.S. et al.: "Familial chronic mucocutaneous candidiasis." J. Med. Genet., 9:302-310, 1972.

CHURG-STRAUSS-ZAK SYNDROME

Synonymy

S. of allergic granulomatosis. Churg-Strauss disease.

Syndromography

Clínic

There are non-specific manifestations: fever, general malaise, anorexia, weight loss.

Pulmonary symptoms predominate due to severe asthmatic attacks, in chronic asthmatic patients, and pulmonary infiltrates.

In 70% of patients there are skin lesions, similar to erythema multiforme, non-thrombocytopenic purpura. Subcutaneous nodules distributed on the trunk and limbs.

Paraclinical examinations

Blood. Anemia. Noticeable eosinophilia of more than 1,000 cells/mm² in more than 80% of patients. Accelerated erythrocyte sedimentation.

Skin and/or muscle biopsy. There is endothelial necrosis, edema, fibrinoid necrosis of collagen fibers and granulomas.

Syndromogenesis and etiology

The exact pathogenesis has not been defined, but hypersensitivity phenomena are suggested.

It is related to vasculitis and collagen diseases.

Literature

Strauss, L.; J. Churg and F.G. Zak: “Cutaneous lesions of allergic granulomatosis: histopathologic study.” J. Invest. Dermatol., 17:349-359, 1951.

Winkelmann, R.K. and W.B. Ditto: "Cutaneous and visceral syndromes of necrotizing of allergic angiitis: a study of 38 cases." Medicine, 43:59-89, 1964.

ISOLATED IgA DEFICIENCY SYNDROME

Synonymy

S. of IgA deficiency. S. of selective gamma-A deficiency.

Syndromography

Clínic

It is the most common immunodeficiency and can begin in infants, at 3 or 4 years of age, or in young adults.

Respiratory infections of varying intensity occur: asthma and atopies.

It has been reported that combined IgE and IgA deficiency did not predispose to recurrent lung infections while IgA-deficient patients with elevated or normal IgE had recurrent lung and sinus infections.

Sometimes patients present chronic diarrhea with steatorrhea due to malabsorption, abdominal pain and sometimes rectal bleeding.

This syndrome has been associated with autoimmune diseases such as lupus and rheumatoid arthritis.

Paraclinical examinations

Immunological study. There is deficiency of IgAIe IgA2 in serum and mucosal secretions.

Precipitating antibodies to IgA have been found.

Determination of IgE

Genetic study. Abnormalities of chromosome 18 are reported.

Syndromogenesis and etiology

This syndrome may be familial, although no single pattern has been observed. It is common for it to present as a sporadic defect.

The cause, whether genetic or due to substances in the environment, leads to a block in the terminal differentiation of B lymphocytes. All patients have B lymphocytes with IgA, although their number may be reduced.

Selective suppression of the IgA response by T cells has been described in some patients. It has been emphasized that many patients have a primary defect in T cell regulatory function.

Patients are at high risk of transfusion reactions.

Bibliography

Ammann, H.R.: "Selective IgA deficiency presentation of 30 cases and a review of the literature." Medicine, 50:223, 1971.

Buser, .; R. Butler and R. Martin du Pan: "Susceptibility to infection. J. Pediat.. 72:153, 1968.

Francois, R.; D. Rosenberg; R. Creyssel, et al.: "Syndrome d'infection a repetition par deficiencies in beta-2-A globulins." Arch. Franc. Pediat., 22:913, 1965.

SELECTIVE IgA DEFICIENCY SYNDROME AND STEATORRHEA

Synonymy

S. of dysgammaglobulinemia with nodular hyperplasia of the intestine.

Syndromography

Clinic

It has been reported in adults. There is steatorrhea, general malaise, bone pain, abdominal distention, and malleolar edema.

Paraclinical examinations

Blood. Anemia. Decrease in IgE values, absence of IgA and IgM.

Radiology. Diffuse lymphoid hyperplasia of the entire intestinal tract. Multiple nodules in mucosa.

Syndromogenesis and etiology

Although affected families have been reported, a defined inheritance pattern has not been demonstrated.

The etiology is unknown.

Literature

Hermans, P.E.; K.A. Huizenga; H.N. Hoffman, et al.: "Dysgamma globulinemia associated with nodular lymphoid hyperplasia of the small intestine." Amer. J. Med., 40:78-89, 1966.

Sell, S.: "Immunological deficiency diseases." Arch. Path., 86:95- -107, 1968.

DYSGAMMAGLOBULINEMIA TYPE I SYNDROME

Syndromography

Clínic

This syndrome is more common in men but is, however, reported in adult women. There is a greater susceptibility to infections.

Paraclinical examinations

Immunoglobulin quantification. Demonstrates decreased levels of IgA; IgM and IgG levels are normal.

Cellular immunology. Normal cellular response.

Syndromogenesis and etiology

The form that affects children may be sex-linked recessive.

The etiology is unknown.

Literature

Sell, S.: Immunological deficiency diseases". Arch. Path., 86:95- -107, 1968.

RETICULAR DYSGENESA SYNDROME

Synonymy

S. of congenital aleucia. S. of panaleucia.

Syndromography

Clínica

There are severe untreatable infections in the first days of life.

Paraclinical examinations

Blood. There is complete agranulocytosis. Only some large lymphocytes and monocytes are observed.

Medulogram. Complete absence of the myeloid series.

Syndromogenesis and etiology

It is characterized by congenital agranulocytosis, lymphopenia, and lymphoid and thymic hypoplasia with absence of humoral and cellular immune functions.

A genetic etiology and autosomal recessive inheritance have been proposed.

Literature

De Vaal, O.M. and V. Seynhaeve: "Reticular dysgenesis." Lancet, 2:1123-1 125, 1959.

HEREDITARY ANGIONEUROTIC EDEMA SYNDROME

Syndromography

Clínica

It usually begins in adolescents and is characterized by skin edema in the form of a circumscribed swelling, with a sudden, deforming appearance, without itching or color changes. It can also appear in the mucous membranes: hypopharynx or larynx, accompanied by hypoxia and danger of death.

When located in the digestive system, it is manifested by abdominal cramps and bilious vomiting due to edema of the intestinal wall; If the colon is affected, diarrhea occurs

watery

Paraclinical examinations

Immunological study. The level of complement inhibitor C1 is only 17% of normal and there is also a decrease in factors C2 and C4.

Syndromogenesis and etiology

Generally there is a considerable deficiency of the C1 fraction of complement in the blood; other times, great increase in an inactive form of the inhibitor, especially in the recessive form of this syndrome.

The etiology is genetic and is transmitted in an autosomal dominant manner, affecting twice as many males as females.

Literature

Landerman, N.S.; M.C. Webster: E.L. Becker, et al.: "Hereditary angioneurotic edema II. Deficiency on inhibitor for serum globulin permeability factor and or plasma kalikrein." J. Allerg., 33:330, 1962.
Landerman, N.S.: "Hereditary angioneurotic edema I. Case reports and review of the literature." J. Allerg., 33:316, 1962.
Osler, W.: "Hereditary angioneurotic edema." Amer. J. Med. Sci., 95:362. 1888.

Stiehm & Fulginti: Immunologic disorders in infants and children.
Bd. Saunders, Philadelphia, p.289, 1973.

FELTY SYNDROME

Synonymy

Felty's disease.

Syndromography

Clínica

There are manifestations of rheumatoid arthritis accompanied by splenomegaly and great susceptibility to infections.

Subcutaneous nodules may be found.

Paraclinical examinations

Blood. Neutropenia, anemia and thrombocytopenia with hypercellular marrow caused by hypersplenism. There are high titers of rheumatoid factor; Circular immune complexes

lantes. Accelerated erythrocyte sedimentation.

Radiology. Secondary osteoporosis.

Syndromogenesis and etiology

It is a chronic process. There is a genetic predisposition.
It is associated with the HLA-DRW4 antigen and has an autoimmune pathogenesis.

There are defects in the function of polymorphonuclear cells. It is accompanied by hypersplenism.

The report of familial cases suggests a genetic etiology.

Literature

Blendis, L.M., et al.: Familial Felty's syndrome". Rheum. Dis.. 35:279-281, 1976.

GITLIN SYNDROME

Synonymy

S. of thymic alymphoplasia. S. de Glanzmann-Riniker.

Syndromography

Clinic

It only affects males from birth. Frequent infections occur.

Paraclinical examinations

Blood. There is lymphopenia of less than 1,000 lymphocytes. Immunoglobulins are decreased.

Immunological study. A failure of the cellular response is demonstrated.

Syndromogenesis and etiology

It is transmitted by sex-linked recessive inheritance.

The etiology is unknown.

Literature

Gitlin, D. and J. Craig: The thymus and other lymphoid tissues in congenital agammaglobulinemia." Pediatrics, 32:517-530, 1963.

EOSINOPHILIC GRANULOMA SYNDROME

Synonymy

S. de Lichtenstein-Jaffe.

Syndromography

Clinic

It affects both sexes and all ages. At first it may be asymptomatic. It is characterized by intermittent low-grade fevers.

Recurrent lymphadenopathy, generally cervical.

Skin manifestations or exophthalmia.

Diabetes insipidus may develop later.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Lichtenstein, L. and H.L. Jaffe: "Eosinophilic granuloma of bone with report of case." Amer. J. Path.. 16:595-604, 1940.

WEGENER GRANULOMATOSIS SYNDROME

Synonymy

S. de Klinger. S. of granulo-arteritis and glomerulonephritis

bushy

Syndromography

Clinic

It occurs in both sexes and in all ages with persistent episodes of rhinitis and sinusitis, pain in the paranasal sinuses, purulent or bloody discharge from the nasal passages and mucosal ulcerations.

There are signs of pulmonary infiltration with symptoms of tracheobronchitis, asthma and pneumonia, cough, hemoptysis and dyspnea.

High blood pressure is rare. In advanced stages, renal failure occurs, arthralgias, polyneuropathy, pericarditis and myocarditis are also described and there is frequently conjunctivitis, episcleritis, scleritis, sclerouveitis.

There are skin lesions (papules, vesicles). palpable purpura, subcutaneous nodules.

Paraclinical examinations

Skin and organ biopsy. Necrotic granulomatous lesions appear, necrotizing vasculitis of arteries and veins. Hyaline and fibrinoid thrombi located in the lungs and kidneys.

Immunofluorescence. There is deposition of IgG in the glomerular basement membrane.

Urine. Albuminuria, cylindruria and hematuria.

Blood. Anemia occurs. Elevated creatinine. Hyperglobulinemia. Transient eosinophilia. Accelerated erythrocyte sedimentation. Increased IgA. Elevated rheumatoid factor.

Syndromogenesis and etiology

It is considered a disease due to immune complex deposition.

The etiology is unknown.

Literature

Klinger, H.: Grenzformen der Periarteritis nodosa". Frankfurt Ztschr. Path., 42: 455-480, 1931.

Stijes, D.P.: "Wegener's granulomatosis" Basic and Clinical Immunology. 5th. ed.,

Toma, G.A.: "Lethal mildine granuloma and Wegener's granulomatosis." J. Laryng., 82:129-139, 1968.

Wegener, E: Uber generlisierte septische gefosser "Krankugen." Verhandt. Deutsch. Path. Gesellsch.. 29:202-210, 1936.

NFANCLA TRANSIENT HYPOGAMMAGLOBULINEMIA SYNDROME

Synonymy

S. of transient hypogammaglobulinemia of the infant.

Syndromography

Clinic

It affects both sexes. At 5 or 6 months of normal life after birth, gastrointestinal disorders, recurrent infections of the respiratory system, and periorbital edema appear.

Paraclinical examinations

Blood. There is anemia. Absence or decrease in IgG, IgM and IgA, which can persist for up to 2 years. Serum IgG at 5 or 6 months of age reaches a minimum value of 350 mg/100 mL.

Syndromogenesis and etiology

Transient deficiency due to a delayed production of immunoglobulins, after the catabolism of maternal IgG acquired transplacentally occurs.

Literature

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IMMUNO COMPLEX SYNDROME

Synonymy

Immune complex disease.

Syndromography

Clinic

Inflammation and tissue damage can arise in an individual from the immune response to foreign antigens or less frequently to autologous components. The immunological reactions that cause pathological processes may involve humoral (antibodies) or cellular immunity.

A consequence of the humoral immune response to antigens is the formation of immune complexes (IC) between the triggering antigens and the specific antibody molecules.

scientific.

These ICs show a wide variety of biological activities. They can suppress or enhance immune responses, inhibit the destruction of tumor cells and deposit on the walls of blood vessels and filtering organs, such as the kidney and others, where tissue damage can occur secondary to the activation of the complement (C) cascade and the attraction of neutrophils and macrophages.

However, IC formation normally protects the host, facilitating the elimination of various antigens and invading microorganisms by the phagocytic system.

It should be noted that the ability of ICs to interfere with immune mechanisms, and to activate or suppress lymphocytic function, constitutes their most notable effect in some diseases.

The fate of circulating ICs is fundamentally determined by the quantitative ratio between antigen and antibody. ICs that form in a large excess of antigens are small in size, do not fix complement, and generally

they cannot initiate inflammatory processes. On the contrary, ICs formed in a significant excess of antibodies, although capable of activating complement, are large and insoluble, being phagocytosed early and, consequently, of limited pathogenicity. The maximum pathogenic potential seems to be between these two extremes of complex size, that is, in ICs formed in moderate antigenic excess. These ICs are intermediate in size and are soluble; They fix complement, but are not rapidly phagocytosed. Consequently, they remain in the circulation and exert their phlogogenic effects with intensity.

The magnitude and duration of antigenic exposure are also important factors. If the antigenic exposure is of short duration, the tissue damage and clinical manifestations are transitory. On the contrary, if the antigen persists in the circulation, or if there is a continuous supply of antigenic material, as occurs in chronic infections or with autoantigens, there is the possibility of prolonged IC formation and these develop the disease.

IC deposition also depends on hemodynamic and anatomical factors. ICs tend to localize in the vessels when there is an increase in blood pressure. Platelets adhere to the endothelium in areas of turbulent flow, and this could explain the greater accumulation of IC at these points. The glomeruli, choroid plexuses, synovium, skin, and uvea share a high rate of blood flow per unit of tissue mass, with the consequent potential for trapping large amounts of IC in their vascular walls; As a consequence, these places have a high risk of presenting disorders related to CIs.

What has been mentioned above helps to understand that there may be different syndromes and diseases due to HF, among which these three include:

1. Serum sickness. There is fever, morbilliform or urticarial eruptions. Arthralgias, less common lymphadenopathy.
   Arthritic manifestations, nephritis, neuropathy and vasculitis.
2. Systemic lupus erythematosus. At first, only one organ or system may be affected. It subsequently affects multiple organs. There is asthenia, general malaise, fever, weight loss.
   Arthralgias, polyarthritis. Malar erythema on "butterfly wings", alopecia. Pale skin and mucous membranes. Splenomegaly, lymphadenopathy. Psychosis, seizures, peripheral neuropathies. Myocardial and pericardial pleural manifestations. Signs of pulmonary hypertension. Venous and arterial thrombosis. Retinal vasculitis. Renal manifestations.
   Vasculitis.
3. Palpable purpura and symptoms similar to those of serum sickness.

Paraclinical examinations

Immunological study. Detection of circulating complexes.

Determination of complement in serum preferably C3 and C4.

Presence of cryoprecipitates. Hepatitis B surface antigen.

Antinuclear antibodies, in particular anti-DNA, anti-Ds DNA.

Immunofluorescence technique.

Syndromogenesis and etiology

Entry of harmful elements with the production of specific antibodies, which are deposited in the tissues.

Causes: Hypersensitivity to drugs. Presence of autoantibodies. Abnormalities in the predominant T lymphocytes

SOres.

IMMUNODEFICIENCY SYNDROME ACCOMPANIED BY SERUM LYMPHOCYTOTOXINS

Syndromography

Clínic

This syndrome has been reported in few patients with recurrent bacterial and fungal infections.

Paraclinical examinations

Blood. Fluctuating lymphopenia. Normal immunoglobulin concentration.

Immunological study. There are alterations in the response of cellular immunity and also of specific antibodies.

Syndromogenesis and etiology

Specific antibodies against p cell antigens cause selective elimination of these cells.

ACQUIRED IMMUNODEFICIENCY SYNDROME (AIDS)

Synonymy

Homosexual pneumonia. S. of lymphadenopathy.

S. symptom complex related to AIDS.

Syndromography

Clinic

It is a serious and fatal clinical disorder that was identified as an independent syndrome in 1981. AIDS represents the last clinical stage of infection by the human immunodeficiency virus (HIV), which is characteristic

main the progressive damage of the immune system with the consequent appearance of opportunistic processes (infections and tumors) and the affectation of other organs and systems of the economy, fundamentally the central nervous system (CNS),

After HIV infection, people may present, over the course of several weeks or months, a self-limited acute infection, similar to infectious mononucleosis, or they may remain asymptomatic for a prolonged period (usually several years) that represents the incubation period, which ends with the appearance of one of the opportunistic infections or tumors that constitute the specific indicators of the underlying immunodeficiency. Opportunistic infections initially included: Pneumocystys carini pneumonia, chronic cryptosporidiosis, CNS toxoplasmosis, esophageal or lower respiratory tract candidiasis, disseminated or CNS cryptococcosis, disseminated atypical mycobacteriosis, and cytomegalovirus pulmonary, gastrointestinal, ocular, or CNS infections, disseminated mucocutaneous herpes simplex virus infections, and leukopathy. progressive multifocal. Cancers include Kaposi sarcoma, primary B-cell lymphoma limited to the brain, and non-Hodgkin lymphoma. Later, in 1987, other diseases that may be indicators were included, including: wasting syndrome, extrapulmonary tuberculosis and dementia or sensory neuropathy due to HIV.

It is possible that in the future new AIDS indicator diseases will be included.

The syndrome can affect any person, regardless of age, sex, ethnic group, nationality, etc., but from an epidemiological point of view, the groups currently at greatest risk are the sexually promiscuous, intravenous drug addicts, and recipients of blood and blood products residing in countries where adequate blood quality control is not carried out, as well as the young children of these people.

From the clinical point of view, an attempt has been made to outline them into different groups or forms of presentation:

Group I. Acute infection, generally asymptomatic, seronegative, but may appear: fever, adenopathy, myalgias and arthralgias, digestive disorders (diarrhea). Headache.

Group II. Asymptomatic infection. It appears after seropositivity.

Group II. Infection called minor AIDS or lymphadenopathy syndrome. Lymphadenopathy is seen in different

Group IV. Manifest illness. One or more opportunistic infectious or tumor-like processes appear.

Examples: Pneumocystis carini, cryptosporidiasis, candidiasis, toxoplasmosis, Kaposi's sarcoma.

Paraclinical examinations

Serology. Since 1985, there has been a diagnostic system to detect antibodies against HIV, and the most commonly used initial detection test is the enzyme-linked immunosorbent assay (ELISA), which is very sensitive and specific.

However, this technique must be complemented with another more specific one such as Western Blot.

It is also possible to determine, by serological techniques, the circulating HIV antigen and nucleotide sequences of the virus by means of the polymerase chain reaction.

Microbiological, histological or other tests.

They are used for confirmation of indicator diseases and are very useful, as is the T helper cell count, which is used to evaluate the intensity of HIV infection.

Blood. Lymphopenia less than 1,500/mm. T4 helper lymphocytes decreased, T8 increased or normal. T4/T8 ratio less than 1. There is a decrease in T function in vivo

(cutaneous anergy). Polyclonal hypergamma IgG-IgA. Determination of antibodies against HTLV/IIILAV (ELISA) Bacteriology. Virus isolation.

Radioimmunoassay.

Syndromogenesis and etiology

The infectious agent is the human immunodeficiency virus (HIV) , a retrovirus of which two types HIV-1 and VH-2 have been identified and which is transmitted by sexual contact, through blood and blood products, and by the maternal-fetal route.

The most accepted mechanism of action is a defect in cellular immunity caused by the HTLV/IIILAV virus, which selectively infects the T lymphocyte subgroup, helpers/inducers (T4).

Sexual contact is the main mechanism of transmission of the virus. Exposure to the virus through penetrating injuries.

The selective defect of the T4 cell causes global defects in different components of immunity. It includes defects in NK (natural killer) cells, cytotoxic T cells with specificity towards viruses, B cells, and monocytes. Defects in the release of interleukins.

The virus can activate B cells, which explains the polyclonal increase in immunoglobulins.

Literature

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IGM ISOLATED IMMUNODEFICIENCY SYNDROME

Syndromography

Clinic

A small percentage of asymptomatic individuals is described. Another 60% had severe recurrent infections often with bacteremia.

Pneumococcus and meningococcus infections have been identified. Gastrointestinal disorders, atopy, splenomegaly, and associated malignant processes may appear.

Paraclinical examinations

Blood. IgM quantification with IgM deficiency. Very low or normal B lymphocytes.

SEVERE COMBINED IMMUNE DEFICIENCY SYNDROME

Syndromography

Clinic

This syndrome is characterized by significant functional damage to humoral and cellular immunity.

Without treatment, infants rarely survive more than a year.

It can present in the form called Swiss-type agammaglobulinemia, as reticular dysgenesis or as an X-linked hereditary model.

There are episodes of repeated infections.

Paraclinical examinations

Determination of lymphocytes. Global count and population count T4, T8 and B.

Immunoglobulin quantification.

Syndromogenesis and etiology

In the Swiss type there is an intense lymphopenia that includes Ty B cells, it is inherited in an autosomal recessive pattern.

In reticular dysgenesis, rarely other hematopoietic cell lines do not develop; The defect resides in the T and B cell precursors.

Half of the patients show deficiency of an enzyme, adenosine deaminase (ADA), which is involved in purine metabolism. Variable degrees of lymphopenia and the deficiency is usually greater in T cells than in B cells (autosomal recessive form).

In the sex-linked hereditary model, there is sometimes no severe lymphopenia and in some cases, few or absent normal B lymphocytes and circulating T lymphocytes are observed. The possibility is suggested that the thymic epithelium is defective or absent.

Some patients have selective T helper (T4) cell deficiency.

KAWASAKI SYNDROME

Synonymy

S. of mucocutaneous lymph nodes.

Syndromography

Clínic

It is an acute syndrome accompanied by fever that affects multiple systems in children. It affects the skin and mucous membranes.

There are lymphadenopathy.

It is benign in nature and disappears spontaneously. It may be associated with a coronary artery aneurysm, in this case with a more unfavorable prognosis due to coronary vasculitis. Aneurysms are identified along the entire length of the coronary arteries.

It is noted that many cases of polyarteritis nodosa reported in children are nothing more than this syndrome accompanied by complications.

Pericarditis, myocarditis, cardiac infarction may occur.

KUSSMAUL-MAIER SYNDROME

Synonymy

S. of polyarteritis nodosa. S. of periarteritis nodosa. Panarteritis nodosa S. S. of hypersensitivity angiitis. S. de Zeek.

Syndromography

Clinic

Around 50% of patients present: fever, weight loss. It affects both sexes.

At first there are vague symptoms such as: discomfort, headache, abdominal pain, myalgia.

In relation to vascular involvement, the following may occur:

Kidney. Pictures of kidney failure and high blood pressure.

Musculoskeletal system. Arthritis, arthralgia, myalgia.

Nervous system. Peripheral neuropathies. Vasculocerebral accidents, seizures.

Gastrointestinal system. Abdominal pain, nausea, vomiting, intestinal infarction and perforation, liver infarction.

Fur. Eruptions, nodules, purpura, livedo reticularis.

Respiratory infections or drug reactions occur.

Fever, general malaise. Weight loss, myalgia, arthralgia, abdominal pain, anginal pain, polyneuritis (with motor and sensory symptoms), hemiplegia, seizures, acute cerebral syndrome, visual disorders.

Asthma episodes, bronchitis.

In the background we find, in the previous months:

respiratory disease (56%), reaction to medications (38%), middle ear infection (31%), high blood pressure (25%).

Physical examination. Skin lesions, polymorphic manifestations: diffuse erythema, purpura, urticaria, ulcerations.

Painful, pulsating nodules in the skin and subcutaneous tissue.

Raynaud's phenomenon. In acute cases there is gangrene.

Arterial hypertension, tachycardia, edema, pericarditis.

Exudate and retinal disorders in the fundus.

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation rate, elevated leukocyte count, hypergammagbulinemia.

Angiography. Aneurysm in the arteries, of fine and medium caliber of kidneys, liver and viscera.

Biopsy of affected organs. Skin and muscles.

Immunological study. Sometimes they are found:

hepatitis B surface antigen, circulatory immune complexes

lantes.

Immunofluorescence. Hepatitis B antigen, IgM and complement in the vessel wall.

Urine. Albuminuria, red blood cells and casts.

Kidney biopsy. It shows the nodules and muscles with the typical changes of this disease.

Syndromogenesis and etiology

It is a multisystem necrotizing vasculitis of fine and medium-caliber arteries with a muscular layer. The muscular layer presents fibrinoid necrosis and decreased caliber with thrombosis. Tissue infarction.

In the classic form there is no involvement of the pulmonary arteries.

Immunological phenomena are pointed out in the pathogenesis of the disease.

The etiology is unknown.

Literature

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WALDENSTRÖM MACROGLOBULINEMIA SYNDROME

Syndromography

Clinic

This syndrome is characterized by the presence of lymphadenopathy and splenomegaly. There is hyperviscosity of the blood with alterations in consciousness. There may be Raynaud's phenomenon.

There are bleeding disorders, hepatomegaly, anemia, recurrent infections, peripheral neuropathies.

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation. M component in serum. Increase in IgM in the serum.

Medulogram. Increase in plasma cells and lymphocytes adults.

SIA test. Positive.

Syndromogenesis and etiology

It affects the bone marrow with excessive production of paraprotein and hyperviscosity.

Cases have been reported in generations of several families and chromosomal abnormality in lymphocytes of an affected family, so a genetic etiology is suggested.

Literature

Mackenzie, M.R.: "Macroglobulinemia an analysis of 40 patients." Blood, 39: 874, 1972.

MYELOPROLIFERATIVE SYNDROME LINKED TO THE X CHROMOSOME

Synonymy

Immunodeficiency V. Susceptibility to Epstein-Bar virus.

Syndromography

Clinic

It is an X-linked recessive syndrome that is accompanied by selective damage to immunity against Epstein-Barr virus (EBV).

When mononucleosis occurs in infected men, the consequences are fulminant and fatal, or it can be accompanied by malignant cell processes, or produce gamma hypoglobulinemia, aplastic anemia, or agranulocytosis.

Paraclinical examinations

Immunological study. Antibodies against EBV have been found in some patients, but immunoglobulins against infections are not detected. A reduction in the activity of killer (NK) cells has been found.

Syndromogenesis and etiology

This syndrome is transmitted by recessive inheritance linked to the X chromosome with selective damage to immunity to the virus. Epstein-Barr and the defect that abolishes the normal response to this type of virus in normal individuals has not been found.

The primary mechanism of EBV infection appears to be the generation of cytotoxic cells and NK cells. They may also function to eliminate virus-infected B cells.

The existence of an X-linked gene that determines susceptibility to acquired immunodeficiency syndrome is suggested.

Heterozygous women show abnormally high titers of antibodies to EBV.

Literature

Purtilo, D.I, et al.: "Hematopathology and pathogenesis of the X-linked recessive lymphoproliferative syndrome." Am. J. Med., 62:225-233, 1977.

PURPLE SYNDROME

DUE TO HYPERSENSITIVITY TO HISTAMINE

Syndromography

Clinic

Painful ecchymoses occur spontaneously.

The injection of histamine produces ecchymosis and systemic manifestations: dyspnea, flushing, nausea, palpitations.

Paraclinical examinations

Blood and urine. Normal levels of serotonin, catecholamines and histamine.

Syndromogenesis and etiology

It is caused by a sensitivity to histamine.

Literature

Schulman, N.R.. J.H. Feigon and T.C. Bithell: "The role of histamine in the pathogenesis of a disease characterized by ecchymosis and systemic symptoms." Clin. Res., 7:216, 1959.

SJÖGREN'S SYNDROME

Synonymy

S. dry. S. de Gougerot-Hower-Sjögren. S. de Gougerot- - Mickuliz-Sjögren. S. of atrophic dacrosialopathy. S. of mucosal disecretosis. S. secret-inhibitor. S. of keratoconjunctivitis sicca.

Syndromography

Clinic

It occurs more frequently in middle age, particularly in women. The disorder arises on its own, and in this case it is known as primary syndrome, or together with other autoimmune diseases and in this situation it is known as secondary. The exact incidence of this syndrome has not been determined, but it is considered that in the United States it is the second most common cause of rheumatic diseases. Even 30% of people with rheumatoid arthritis, 10% with systemic lupus erythematosus, and 1% of patients with scleroderma show secondary Sjögren's syndrome.

The most common manifestations are: keratoconjunctivitis sicca and xerostomia.

Patients initially experience a sensation of sand in their eyes and dry mouth. Dry mucous membranes can extend to the upper respiratory tract, with persistent cough and hoarseness. Dryness of the conea can cause ulcerations.

In cases of primary syndrome there is kidney involvement (minimal interstitial nephritis). It rarely causes chronic kidney failure. A small number of patients with nephropathy show immune complex glomerulonephritis.

25% of patients may be accompanied by vasculitis that manifests itself in the form of cutaneous purpura or vasculitis due to hypersensitivity of the lower limbs, or severe vasculitis and cryoglobulinemia causing fever, skin rashes, and intestinal infarctions.

Neurological manifestations may occur (sensory polyneuropathy, mononeuritis), as well as diffuse proximal myositis.

The picture of pseudolymphoma is observed: with inphadenopathy, parotid enlargement or lung nodules, autoimmune thyroiditis with signs of biochemical hypothyroidism.

Polyarthritis, premature tooth destruction, and dryness of the skin and vagina are not uncommon.

Other abnormalities occur: lymphoproliferative neoplasms.

Paraclinical examinations

Blood. Circulating immune complexes. Autoantibodies. Leukopenia, accelerated erythrocyte sedimentation rate, thrombocytosis.

Immunological study. Increased antiglobulin antibodies, antinuclear antibodies, high concentrations of gamma globulin. Circulating immunocomplexes.

Sialography. Saliectasia, destruction and atrophy of the salivary ducts.

Chest radiology. Reticulonodular infiltrate. Hilar nodular thickening. Bronchiectasis.

Lymphography. Nodular thickening with foamy reticulum pattern. Juxtarticular destruction. Nephrocalcinosis. Atrophy of the esophageal mucosa.

Syndromogenesis and etiology

It is an immunological disorder characterized by the progressive destruction of exocrine glands accompanied by various immunological phenomena.

There is a primary form and a secondary form where other autoimmune diseases are associated.

Two types of immunoregulation defects are found:

1. An active abnormality in the cellular immunity system with inflammatory infiltrates consisting of activated T lymphocytes and B lymphocytes.
2. Activation of B lymphocytes with monoclonal hypergammaglobulinemia.

The two main mechanisms of tissue destruction in this syndrome are infiltration by lymphocytes and deposition of immune complexes.

Immunogenetic predisposition appears to play an important role in the incidence of the syndrome, and the frequency of histocompatibility antigens HLA-B8, HLA-DRW3 and MT-2 is significantly higher in patients with the primary syndrome.

Literature

Deutsch, HJ.: "Sjögren's syndrome and pseudolymphoma." Ann. Otol.. 76:1075-1084, 1967.

Fox, R.I., et al.: "Primary Sjögrens syndrome: cynical and immunopathological features." Semin. Arthritis Rheum., 14:77, 1984.

Sjögren, H.: "Zur Kenntis der Keratoconjunctivitis sicca (Keratitis filiformis bei Hypofunktion der Tramendrusen)". Acta Ophth. (Supp. II), 1-151, 1933.

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POLYANGIITIS OVERLAP SYNDROME

Syndromography

Clinic

There is overlapping of symptoms of classic polyarteritis nodosa, angitis, and allergic granulomatosis.

Manifestations of the vasculitis group due to hypersensitivity of small vessels also occur.

This subgroup has been recognized as overlapping polyangiitis syndrome, and is part of a larger group of systemic necrotizing vasculitis.

This syndrome has been individualized to avoid confusion in attempts to include this overlap syndrome in one of the more classic vasculitis syndromes.

TAKAYASU SYNDROME

Synonymy

S. de Martorell II. S. de Raeder-Arbitz. S. of reverse coarctation. Pulseless disease. S. of chronic obstruction of the subclavian artery. S. of brachiocephalic obliterans arteritis. S. of the aortic arch in young women (see aortic arch syndrome).

S. of idiopathic panarteritis. S. arteritis of large arteries.

Takayasu arteritis.

Syndromography

Clínic

It is a systemic disease with generalized and localized manifestations. It occurs more frequently in young women 10:1. There is discomfort, fever, arthralgia, weight loss, night sweats (symptoms that precede vascular injury).

Generally there is transient unilateral amblyopia or persistent blindness, aphasia, transient hemiparesis, headache, vertigo, syncope, seizures. Weakness and pain in the chewing muscles. Occasionally weakness, numbness or pain during effort in the upper limbs.

Pain over the affected vessels, later accompanied by episodes of organ ischemia. There is an absence of pulses, particularly from the subclavian artery. Arterial murmurs, arterial hypertension. Heart failure secondary to aortic or pulmonary hypertension may occur. In addition, there are syncopal crises and ophthalmological symptoms.

Physical examination. Atrophy and pigmentation of the skin of the face, ulcerations of the nose and soft palate. Temporary amaurosis, eventually retinal atrophy. Cataracts.

Fundus of the eye: anastomosis around the papilla, atrophy or pigmentation of the retina, atrophy of the iris.

Weakness of the pulse in the arms with a drop in blood pressure, a tendency to hypertension in the legs.

Murmurs in the neck and upper chest region. Peripheral gangrene.

Insufficient irrigation of the cerebellum that causes vertigo, hyperexcitability, memory disorders.

Paraclinical examinations

Blood. Normal or altered erythrocytes and leukocytes, accelerated erythrocyte sedimentation, hypergammaglobulinemia.

Chest radiology. Notching of the upper ribs, occasionally calcification of the aotic arch.

Aortography. Partial or total systemic arterial obstruction, extensive collateral circulation, arterial calcifications.

Digital subtraction arteriography. Reveals stenosis or occlusion.

Syndromogenesis and etiology

It is characterized by being an inflammatory and stenotic disease of medium and large caliber arteries that affects the aortic arch and its branches, less frequently the pulmonary one.

It is associated with HLA-DR2 antigens in Japan and HLA- -OR4 in the USA.

Partial occlusion of the renal artery is common.

It is considered an idiopathic arteritis, possibly autoimmune.

Occasional association with: idiopathic ulcerative colitis, ankylosing spondylitis, rheumatoid arthritis.

The etiology is unknown.

Literature

Domingo, R.T.; T.P. Maramba; L.F. Torres, et al.: “Acquired aorto-arteritis: a worldwide yascular entity", Arch. Surg.. 95:780- -790, 1967.

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WISKOTT-ALDRICH SYNDROME

Synonymy

S. de Aldrich. S. de Dees-Aldrich. S. of eczema-thrombocytopenia-repeated infections.

Syndromography

Clinic

This syndrome occurs in infancy and childhood. It only affects men.

It is characterized by eczema, which manifests itself at one year of age.

Recurrent infections, especially otitis media.

Bloody diarrhea, petechiae and hematomas.

Children usually have hemorrhages during infancy and most die from them, infections or lymphoreticular malignancies.

Paraclinical examinations

Blood. There is anemia, thrombocytopenia, lymphopenia. Low levels of IgM. Normal IgG. IgAe IgE increased.

Immunological study. Normal number of B cells. Normal T lymphocyte function.

Medulogram. Normal megakaryocytes. Reduced production of

Syndromogenesis and etiology

This syndrome is transmitted by recessive inheritance linked to the

It has been shown that there are no surface glycoproteins on lymphocytes and platelets.

The etiology is genetic.

Literature

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RELATED IMMUNE SYNDROMES

Syndromes with interstitial calcinosis

Circumscribed calcinosis.

Universal calcinosis.

Interstitial calcinosis may be associated with these syndromes:

Rheumatoid arthritis.

Burnett.

Dermatomyositis.

Scleroderma.

Hyperparathyroidism.

Hypoparathyroidism.

Parasitic infestation.

Kidney failure.

Vitamin D poisoning.

Lupus erythematosus.

Pseudohypoparathyroidism.

Raynaud.

Immune deficiency syndromes

PRIMARY

Swiss type agammaglobulinemia.

Pharyngeal pouch.

Bruton type agammaglobulinemia.

Selective IgA deficiency-steatorrhea.

Say George.

Dysgammaglobulin Type I.

Dysgammaglobulin Type II.

Reticular dysgenesis.

Immune deficiency with hypergammaglobulinemia.

Short stature-high IgA-asthma.

Glanzmann-Riniker.

Gitlin,

Hypergammaglobulinemia-recurrent infections.

Transient hypoglobulinemia in childhood.

Louis-Barr.

Nezelof.

Wiskott-Aldrich.

SECONDARY

Heavy chains.

Hodgkin.

Kahler-Bozzolo.

Myeloma.

Other lymphomas including lymphatic leukemia.

Posttreatment with cytostatics, immunosuppressants.

Waldenstrom.

Thymic tumor syndromes

Erythrocyte aplasia-malabsorption.

Erb-Goldflam.

Thymoma-hemolytic anemia.

Thymoma-rheumatoid arthritis.

Thymoma-hematopoietic insufficiency.

Thymoma-systemic lupus erythematosus.

Thymoma-myocarditis.

Thymoma-plasma cell myeloma.

Thymoma-Cushing syndrome (see ectopic production of hormones due to neoplasms).

Thymoma-Sjögren's syndrome.

Syndromes with necrotizing vasculitis

Bazin.

Churg-Strauss-Zak.

Degos.

Acute febrile neutrophilic dermatosis.

Erythema elevatum diutinum.

Erythema nodosum.

Lethal midline granuloma.

Wegener's granulomatosis:

Horton.

Kussmaul-Maier.

Lindberg.

Löfgren.

Schönlein-Henoch.

Cutaneous-intestinal-oropharyngeal ulcerations.

Urbach.

Allergic vasculitis.

Vilanova-Aguade.

Whitfeld.

Zeek.

HEAT SYNDROME

Synonymy

Heat cramps. Heat exhaustion. Heat injury. Heatstroke.

Syndromography

Clínica

The frequency of heat syndromes is unknown, but in any given summer there may be several hundred in some countries. Particularly susceptible are elderly people, alcoholics and mentally ill people, or those who are under antipsychotic treatment or with diuretics and anticholinergics.
Heat syndromes are especially common during the first days of a heat wave or wave, before a period of adaptation to it occurs.

Heat cramps are the most benign syndrome, and are characterized by painful spasms of voluntary muscles and generally occur after strenuous exercise, even if the environmental temperature is not higher than body temperature. Exhaustion, prostration and heat collapse are the most common manifestations and occur mainly in the elderly. They are sudden and include weakness, nausea, vomiting, urgency to defecate and fainting. During the acute phase, the patient appears grayish in color and the skin is cold and slimy, the pupils are dilated, and the blood pressure may be decreased and the pulse accelerated.

The syndrome can also occur in individuals who perform intense physical exercise in an environment with high temperatures when there is a lot of humidity.

Finally, there is the so-called heat stroke, hyperpyrexia or heatstroke, which is also more common in the elderly with pre-existing chronic illnesses, recruits or long-distance runners. In these cases, loss of consciousness is most common; there may also be headaches, vertigo, fainting, confusion and hyperpnea.

Paraclinical examinations

Blood. Hemoconcentration, elevation of blood urea, rhombocytopenia, prolongation of bleeding and prothrombin time.

Urine. Proteinuria and cylindruria.

Electrocardiogram. Tachycardia, sinus arrhythmia, T wave flattening and inversion, and ST segment depression.

Syndromogenesis and etiology

The syndrome occurs as a consequence of exposure to high temperature and humidity, when the body's adaptation mechanisms fail.

Literature

Britt, B.A.: "Etiology of malignant hyperthermia." Fed. Proc., 38:44. 1979.

Clowes, Jr. G.H.A. and T.E. 0'Donnel, Jr.: “Current concepts: Heat stroke". N. Engl. J. Med., 291:564, 1974.

Costrini, A.M. et al. : "Cardiovascular and metabolic manifestations of heat stroke and severe heat examination." Am. J. Med., 66:296, 1979.

COWDEN SYNDROME

Syndromography

Clinic

The syndrome is characteristic of young women. A hypertrophy (virginal) of the breasts is observed, a nodular cystic breast with thick nodules, with hyperpigmented skin on the breast. It is accompanied by a fascie that is described as birdlike, with mandibular hypoplasia, elevated palatine arch, scrotal tongue, and papillomatosis of the lips and oral mucosa. Dental anomalies with early tooth loss, due to gingival fibromatosis. Repeated sinusitis and rhinopharyngitis. Transmission hearing loss. The association with thyroid adenomas, scoliosis and pectus excavatum is common.

This syndrome represents a variety of gingival fibromatosis-mammary fibromatosis.

Paraclinical examinations

Biopsy of a wart. Demonstrates papillomatous type lesions.

Syndromogenesis and etiology

The etiology is unknown, it is probably a disorder of genetic origin.

Literature

Lloyd, K.M. and M. Dennis: "Cowden's disease, a possible new symptom complex with multiple system involvement." Ann. Intern. Med., 58:136-142, 1963.

INTENSIVE CARE SYNDROME

Syndromography

Clinic

It is an acute behavioral disorder that occurs in patients who have been subjected in an intense and prolonged manner to a wide variety of therapeutic and diagnostic procedures (monitoring, intravenous infusions, subclavian catheters, etc.) and contact with other patients in the same or worse situation. This syndrome is seen more frequently in patients admitted to coronary care units.

Paraclinical examinations

Electroencephalogram, computed axial tomography and three special tests. Using these tests, the diagnosis is made to rule out organic neurological lesions.

Syndromogenesis and etiology

Individual inability to control the various stress situations to which these patients are subjected.

Literature

McKegney, FP.: "The intensive care syndrome." Conn. Med. 30:633-639, 1966.
Nahum, L.H.: “Madness in the recovery room from open heart surgery or "They kept waking me up." Conn. Med., 29:771-772, 1965.

DEMARQUAY SYNDROME

Synonymy

S. fistula of the lower lip and cleft palate.

Syndromography

Clínic

There is no preference for any sex and their frequency of appearance is very low, around 1:75,000 or 1:100,000. Lower lip fistulas appear as red pits or depressions located equidistant from the midline, although occasionally there may be a certain degree of asymmetry. The depressions also have different degrees of depth and are generally asymptomatic or may secrete small amounts of saliva. Some family members may have a double lower lip. Other associated malformations that may be found are: genitourinary and limb abnormalities.

Paraclinical examinations

Biopsy of the lesion. Stratified squamous epithelium. Large epithelial cells with small nuclei can also be found.

Syndromogenesis and etiology

It is transmitted by inheritance, autosomal dominant with a high degree of penetration.

The etiology is unknown.

Literature

Cervenka, J.; R.J. Gorlin and V.E. Anderson: "The syndrome of pits of the lower lip and cleft lip and/or palate, genetic considerations." Amer. J. Human Genet., 19:416-432, 1967.
Demarquay, J.N.: "Quelques considerations sur le bec-de-lievre." Gaz. Med. Paris, 13:52-53, 1845.

DENTINOGENESIS IMPERFECTA SYNDROME

Synonymy

S. with Carmelite teeth. S. de Cadepont. S. of opalescent dentine.

Syndromography

Clínic

It can be observed in people of both sexes. The teeth are rather small in size, orange or brownish and softly translucent. Almost all or a large part of the teeth wear down progressively to the level of the gums. This appearance can be observed in both temporary and permanent teeth.

In some cases it is associated with osteogenesis imperfecta.

Paraclinical examinations

Blood. Everything is normal, except for phosphates, which are elevated in some cases.

Radiology. Anomalies can be observed in teeth or in a bone survey.

Histology. The dentin structure is affected with characteristic histological changes. There is loss of strength with a non-tubular characteristic of the dentin.

Syndromogenesis and etiology

It is an autosomal dominant inherited disease.

The etiology is unknown.

Literature

Stainton, C.W.: "Crownsless teeth. Dental Cosmos, 24:972, 1892.
Ellner, R.: “Die Klinik und Morphologie der capdepontischen Erkrankung.” Denth. Zahnaarzt Stschr., 15:479-486, 1960.

DISASTER SYNDROME

Syndromography

Clinic

It is a name applicable to groups of people or communities that have suffered a disaster and that almost always adopt similar characteristics after it.

The following phases can be considered:

First phase. It occurs a few minutes or hours after the disaster: daze, apathy, disorganized behavior, they do not respond in a coordinated manner.
Second phase. It can last a few days: increased suggestibility, they try to help themselves, but with reduced efficiency, they minimize their own care needs. At first they feel guilty because they have survived and could not help others.
Third phase. After a few weeks they are seen with moderate euphoria, enthusiasm for rebuilding and repairing the damage, and a feeling of communication with the environment that surrounds them.
Fourth phase. Gradual loss of the previous characteristics until their disappearance. Hypercriticism and pity then appear.

Syndromogenesis and etiology

The triggering cause of this syndrome is a large de-

tailor.

Literature

Garb, S: Disaster Handbook. New York, Springer, 1964.

BONE DEMINERALIZATION SYNDROME IN ASTRONAUTS

Significant bone demineralization related to diet and immobilization is observed during space flights.

forced tion produced by confinement in the spacecraft.
Diet rich in calcium and exercise reduce demineralization.

Bibliography

Mack, P.B.; P.A LaChance: GP. Vose et al.: "Bone demineralization of foot and hand of Gemini-Titan IV, V, and VII astronauts during orbital flight." Amer. J. Roentgenol., 100:503-511, 1967.

GENERAL MALIGNANT SYNDROME

Synonymy

S. toxic. S. of anorexia-cachexia.

Syndromography

Clínica

It is characterized by the joint appearance and not attributable to known causes of asthenia, anorexia and weight loss. It is considered that weight loss must be greater than 5% of usual in the last six months. This is an objective fact verifiable by the patient and the doctor, but asthenia and anorexia depend on the subjective opinion of the patient in terms of measuring their intensity. In other patients, nausea and vomiting may appear, as well as amenorrhea in women.

Paraclinical examinations

Blood. Leukocytosis, iron deficiency anemia. Erythrose-elevated sedimentation. Tendency to metabolic acidosis. Decrease in proteins. Negative nitrogen balance.

Syndromogenesis and etiology

Different mechanisms have been proposed to explain this syndrome:

1. That there are circulating anorexigenic peptides, or effects of a tumor on the appetite centers.
2. Modern studies (always in the field of cancer) suggest that it may be a newly discovered metabolite "caquectin", a protein derived from macrophages, similar to "tumor growth factor". At present, it has been shown that both proteins are identical, that they have been found in much greater quantities in cancer patients than in healthy subjects, and that they would explain the three symptoms reported in the clinic. It remains to be demonstrated whether these proteins are present in other pathologies in which this syndrome also exists.

In its etiology, different processes are indicated in which the syndrome is present: cancer of different types (which is why it has been considered within the paraneoplastic syndromes), gastrointestinal diseases (malabsorption, chronic inflammatory bowel diseases, diabetic enteropathy), fever of unknown origin (tuberculosis, sarcoidosis, brucellosis, collagen diseases such as giant cell arteritis and vasculitis).

Literature

Soriano, M.: The common toxic syndrome", Med. Clin.. 70:425- -431, 1978.
Theologides, A.: Pathogenesis of cachexia in cancer.
A review and hypothesis". Cancer, 29:484-488, 1972.
Theologides, A.: "The anorexia-cachexia syndrome: a new hypothesis." Ann. N.Y. Acad. Sci., 230: 14-22, 1974.

EXPANSIVE BLOW SYNDROME

Synonymy

S. of detonation injury.

Syndromography

Clínic

Symptoms include dyspnea, ear pain, partial or complete deafness, and pain in the chest or other region, accompanied by loss of consciousness.

Physical examination. Shock, bradycardia and signs of damage to the heart, lungs and abdominal organs. There may be abdominal distention and petechiae.

The severity of the injuries causes death in many cases. The autopsy shows hemorrhages in the lung parenchyma, emboli in arterioles and pneumothorax.

Paraclinical examinations

Electrocardiogram. Atrial fibrillation.

Chest radiology. Pleuropulmonary injuries.

Syndromogenesis and etiology

The clinical picture occurs as a result of the high or low pressure phases that occur after detonations.

Literature

De Candole, C.A.: "Blast injury." Dog. Med. Ass. J., 96:207-2 14, 1967.

GULF WAR SYNDROME

Syndromography

Clinic

This syndrome has been described in hundreds of English and North American veterans who participated in the Gulf War against Iraq (1990-1991).

The main symptoms are: fatigue, tiredness, muscle pain, headache or hemicrania, dizziness, unexplained memory loss.

They were present in thousands of soldiers in whole or in part.

A high incidence of children born with malformations has also been detected in these combatants after their return.

The detailed examination of the conditions of the nervous system of these soldiers revealed the existence of obvious damage to the nerve endings of the members.

Syndromogenesis and etiology

According to the team at the Southern General Hospital in Glasgow, Scotland, led by Jamal, neurological damage must be attributed to contact, inhalation or ingestion of chemical substances and more precisely "organic compounds." The nerves show damage similar to that suffered by sheep covered with antiparasitic substances based on organophosphates or raised on land treated with fertilizer based on these compounds, which in chemical profile closely resemble nervine gas, one of the most fearsome chemical-biological weapons produced by man.

It is known that the Allied troops in that war were given organophosphates in pill form to protect them from possible nerve gas attacks.

The same compounds were part of the insecticides that soldiers used abundantly to keep insects and desert parasites away from tents and camps, and avoid diseases such as malaria, leishmaniasis and others.

Researchers at Duke University in the US established that the mixture of two pesticides (Deet and Permethrine) and an antidote for gases (pyridostigmine bromide), used by soldiers to protect themselves from mustard gas, caused neurological reactions similar to those exhibited by ex-combatants.

The study concludes that the weak absorption of just one of these products is not dangerous, but the mixture of all three is very toxic.

Leishmaniasis, which some believed could cause the symptoms, was only present in 32 of the 19,000 soldiers examined.

Literature

Jamal, Ga.; S. Hansen; "F. Apartapoulos and A. Pedem: The Gulf War".

Syndrome". Is there evidence of dysfunction in the Nervous system'".
J. Neurol. Neurosurg. Psychiat., 60(4):449-451, 1996.

MALIGNANT HYPERTHERMIA SYNDROME

Syndromography

Clinic

The presence of this syndrome may be suspected if relaxation occurs during the induction of anesthesia, or if muscle fasciculations become evident when succinylcholine is administered. The presence of lockjaw may be the first manifestation in some patients. There is then a very rapid rise in temperature. Hot skin followed by tachycardia and cardiac arrhythmia, hypotension and mottled cyanosis is also an early sign.

Paraclinical examinations

Blood. Hyperkalemia and hypermagnesemia, elevations in pyruvate and lactate.

Syndromogenesis and etiology

This is a group of hereditary disorders that manifest after inhalation of anesthetics such as halothane, methoxyflurane, cyclopropane and ethyl ether, or muscle relaxants such as succinylcholine.

Literature

Gronert, G.A.: "Malignant hyperthermia." Anesthesiology, 53:395, 1980.

McPherson, E.W; AC Taylor, Jr.: "The King syndrome: Malignant hyperthermia myopathy and multiple anomalies." Am. J. Med.
Genet., 8:159, 1981.

HPERVENTILATION SYNDROME

Syndromography

Clinic

This syndrome is more common in women and accompanies anxiety or stress problems.

It is characterized by sighing, deep, sometimes unconscious breathing or polypnea. This is followed by vague sensations such as tingling and coldness with paresthesias in the limbs.

Sensation of numbness and dizziness that can lead to loss of consciousness and sometimes palpitations. Anxiety or anguish, sweating of hands and armpits. Some cases may have carpopedal spasms. If consciousness is lost, tonic contractions may occur and the diagnosis may be confused with an epileptic syndrome.

Syndromogenesis and etiology

The clinical picture is due to an excessive elimination of CO, due to hyperventilation, which causes a decrease in PCO in the blood and alkalosis; It is frequently accompanied by transient hypocalcemia that contributes to carpopedal spasms.

If these patients are made to breathe deeply and/or quickly outside of seizures, for 3-4 minutes, the symptoms reproduce, allowing diagnosis. If, on the other hand, we make him breathe in a plastic bag without removing his mouth from the bag, the symptoms disappear.

MONGE SYNDROME

Synonymy

S. chronic of the mountaineers. S. del soroche. Mountain sickness. S. of hypobaropathy. S. de d'Acosta.

Syndromography

Clínic

Two types are described: emphysematous and erythremic. The emphysematous type is characterized by dyspnea, frequent bronchitis and laryngitis accompanied by cyanosis.

In the erythremic type, the predominant symptoms are: fatigue, dyspnea on exertion, mental dullness, headache, epistaxis, hemoptysis, gingival hemorrhage, anorexia, nausea, vomiting, decreased visual acuity, tinnitus, and cough. There is also loss of libido, paresthesia and pain in the limbs, aphonia and lethargy that can progress, in some cases, to coma.

In general, the clinical manifestations can be grouped into:

1. Respiratory. Dyspnea, tachypnea, sometimes Cheyne-Stokes respiration, respiratory cyanosis.
2. Psychics. Lack of will, indifference, euphoria, depression.
3. Neuromuscular. Coordination disorders, muscle and limb heaviness, tremors, vertigo, hyperkinesia, tonic-clonic cramps; sometimes paralysis and loss of consciousness.
4. Visuals. Hemeralopia, decreased ability to distinguish colors.
5. Auditory. Perceptual deafness.

Physical examination. The most frequently found signs are: cyanosis (which increases with exercise), purpura, eyelid edema, bluish injection of the sclera, thickening of the tongue, clubbed fingers. 10% have hepatosplenomegaly. The remission of the condition is the most frequently observed, its total disappearance is achieved by taking the patient from the mountainous area to the plains or to sea level.

Death can sometimes occur due to hemorrhage and bronchopneumonia.

Paraclinical examinations

Blood. Erythrocytosis, reticulocytes are normal or increased. Polyglobulia in people who spend a long time at heights. Leukocytes and platelets are normal. An increase in indirect bilirubin and hematocrit is observed.

Urine. Increased urobilinogen.

Syndromogenesis and etiology

This syndrome is very common in Peru, Bolivia and nations with highlands, of course for people who do not live stably in those places and have not had time to adapt.
be up to par.

The cause of this syndrome is polycythemia secondary to alterations in oxygen pressures that occur in places located at high altitudes above sea level.

It is produced by a decrease in Oxygen tension, which leads to less arterialization of the blood with consequent hypoxia for the tissues, especially in chronic cases.

Symptoms, especially neurological symptoms, are due to cerebral hypoxemia aggravated by compensatory hyperventilation.

Literature

d'Acosta, J.: "Strange effect that the air or wind that flows has in Indian lands." Forums lib. 3 Ch. 9: Natural and moral history of the Indies. Seville. Juan de León, 1950.

Monge, C.: "High altitude disease." Intern Arch. Med., 59:32- -40,1937.

Monge, C.; R. Lozano and A. Carcelen: "Renal excretion of bicarbonate in high altitude natives and in natives with chronic mountain sickness." J. Clin. Invest., 43:2303-2309, 1964.

PLAGIOCEFALY SYNDROME

Synonymy

S. of facial asymmetry. S. of alteration of the dental articulation. S. of facial malformation and asymmetry of the skull base.

Syndromography

Clínic

The condition is characterized by malformation of the base and vault of the skull and disturbances in the dental articulation. Sometimes there may be some mandibular compensation that reduces the joint problems of different degrees of malocclusion: it may be associated with various degrees of plagiocephaly.
The severity of the condition can be very variable.

Syndromogenesis and etiology

It is a congenital malformation, in which hereditary transmission is considered.

Literature

Delaire, J.; J. Billet; J.C. Ferre et al.: "Facial malformations et asymmetrie de la base du crane." Rev. Stomat., 66:379-396, 1965.

Korkhaus, G.: *Línfluence de l'heredité et du milieu sur lárchitecture du crane facial". Rev. Belge Sci. Dentair., 385-403, 1952.

ACUTE RADIATION SYNDROME

Syndromography

Clínic

The clinical manifestations produced by acute radiation exposure are characterized by a temporosequence evolution and also by different clinical forms of presentation (cerebral, gastrointestinal and hematopoietic), which depend on the amount of radiation received and the individual susceptibility of the patient. The typical syndrome has the following characteristics:

1. Prodromal period. It appears within the first two hours after exposure and is characterized by nausea, anorexia, general malaise and fatigue. The symptoms progress towards prostration. On the second day, occasional nausea and vomiting may be the main symptoms.
2. Latency stage. On the third day the patient may be asymptomatic and is able to return to his usual activities, remaining this way for approximately 20 days. If the dose was greater than 300 r, hair loss may begin in the second week.
3. Stage of spinal depression. Anemia, leukopenia and thrombocytopenia appear. Acute onset illnesses, fatigue, dyspnea on exertion, hemorrhages, purpura, stomatitis and fever. This phase or stage lasts approximately 10 more days. In total approximately 30 days.
4. Recovery phase. If the patient manages to survive, progressive recovery and the gradual disappearance of symptoms occur in another month. The convalescent stage begins at 60 days and is completed in 90 days.

Paraclinical examinations

Blood. Anemia, leukopenia and thrombocytopenia.
Hypoproteinemia
Urine. Albuminuria, tyrosinuria.
Medulogram. The symptoms of spinal cord aplasia or tumor lesions of the hemolymphopoietic system are characteristic.

Syndromogenesis and etiology

It is due to the exposure and penetration of gamma rays or neutron radiation into the body. The effects on the person and the evolution are in direct relation to the dose received.

Literature

Gerstner, H.B.: Acute radiation syndrome in man; military and civil defense aspects". U.S. Armed Forces Med. J. 9:313-354.
1958.

SWEAT RETENTION SYNDROME

Synonymy

S. of tropical anhidrotic asthenia. S. of thermogenic anhidrosis. S. of hypohidrosis.

Clinic

The symptomatic picture appears when people affected by the syndrome are exposed to temperature and

high humidity, and is characterized by dizziness, nausea, flushing, pruritus and headache, accompanied by tachycardia, tachypnea and fever. Sometimes, if hyperthermia is severe, the consequences can be serious for the patient.

Paraclinical examinations

Colorimetric tests. You can check the absence of sweating.

Syndromogenesis and etiology

The clinical picture can occur in any of the causes that prevent sweating, such as: congenital absence of sweat glands, or their functional deficit, dermatitis, ectodermal dysplasia, neurological conditions, miliaria and endocrine disorders.

Literature

Shelley, W.B.: P.N. Horvath and D.M. Pillsbury: Anhidrosis; etiologic interpretation." Medicine, 29:195-224, 1950.

Wolkin, J.; J.I. Goodman and W.E. Kelley: "Failure of sweat mechanism in desert." J. A.M.A., 124:478-482, 1944.

SWINGING MOVEMENT SENSITIVITY SYNDROME

Synonymy

S. of neuropathy to movement.

Syndromography

Clínic

It is observed in some people when they travel in cars, trains, boats or airplanes and is characterized by paleness, sweating, salivation, nausea and vomiting; If conditions persist, prostration may ensue.

Syndromogenesis and etiology

The etiology is not defined, it is probably due to the combination of labyrinthine stimulation due to movement and physical, visual or olfactory factors, which may have a stimulating role.

Literature

Kabat, H. and A. Saltz: "Electromyography- aid to diagnosis of amyothrophic lateral sclerosis and motor root compression syndrome." R.I. Med. J., 41:617-620, 1958.

RELATED MISCELLANEOUS SYNDROMES

Syndromes with hereditary cancer

Ancell.

Bourneville.

Basal cell carcinoma.

Chediak-Higashi.

From Sanctis-Cacchione.

Gardner.

Peutz-Jeghers.

Familial adenomatous polyposis of the colon.

Retinoblastoma.

Sipple.

Tylosis carcinoma of the esophagus.

Von Recklinghausen.

Wermer.

Humoral syndromes associated with non-endocrine tumors

Harris.

Hypercalcemia in neoplasms.

Lambert-Eaton.

Polycythemia associated with tumors.

Ectopic ACTH production.

Ectopic gonadotropin production.

Ectopic TSH production.

Inappropriate antidiuretic hormone production.

Syndromes with macroglossia

Glycogen storage.

Amyloidosis.

Congenital hypothyroidism.

Lymphangioma and/or hemangioma.

Neurofibromatosis.

Pseudomacroglossia in Mongoloids.

Wiedemann.

“Mega” syndromes

Cerebral and ventricular.

Hydrocephalus.

Mega" bladder.

Megaloureter.

Megalopelvis (hydronephrosis).

Somatic.

Hemihypertrophy.

Gastrointestinal tract.

Megaesophagus

Idiopathic dilation of the esophagus, cardiospasm.

Megastomach.

Gastric dilation.

Pyloric hypertrophy.

Megacolon.

Segmental dilation of the colon.

Pseudo-Hirschsprung.

Syndromes with head and neck chemodectomas

Paraganglioma (non-chromaffin) of the neck.

Carotid body tumor.

Glomus jugulare tumor.

Vagal node tumor.

Radiation syndromes

Sharp.

Cardiac.

Cerebral.

Radiation fibromatosis.

Liver.

Post-radiation vascular insufficiency.

ACHONDROPLASIA SYNDROME
INCOMPLETE REGIONAL WITH ABDOMINAL MUSCULAR DYSPLASIA

Syndromography

Clinic

There is regional achondroplasia of the ribs and associated ilium.
caused by weakness of the abdominal muscles.

Paraclinical examinations

Radiology. Bone survey that reveals the abnormalities
malias

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance with incomplete

The etiology is unknown.

Literature

Shapira, E.; E. Fischel; S. Moses et al.: "Syndrome of incomplete regional achondroplasia (ilumand ribs) with abdominal muscle dysplasia." Arch. Dis. Child., 40:694-697, 1965.

HEREDITARY PERIODIC DYNAMIC SYNDROME

Synonymy

S. of Eulenburg's hyperkalemic periodic paralysis. S. of paramyotonia congenita. S. of hereditary episodic adynamia.

Syndromography

Clínica

This syndrome occurs in both sexes and is more severe in men. It is installed in the first ten years of life.

It is characterized by attacks during which the patient reports muscle weakness, generally localized in a single muscle group. It can affect the muscles of the face.

There are paresthesias and cramps lasting an hour or more, hyporeflexia and myotonia localized in the tongue or in the lips and eyelids.

The attacks occur during the day. The patient is symptom-free between attacks. There are precipitating factors such as exercise, cold and hunger.

Paraclinical examinations

Blood. Elevated serum potassium during attacks. The administration of l cc potassium induces the crisis.

Syndromogenesis and etiology

It is an autosomal dominant inherited condition. The pathogenic mechanism seems to be the reduction of the muscle membrane potential due to an alteration of the protein gene (alpha subunit), which forms the sodium channels of skeletal muscle. The location of the gene has been assigned to chromosome 17q23.1-q25.3.

Literature

Eulenberg, A.: "Ueber eine familiare, durch 6 Generationen Verfolgbare. Form Kongenitaler Paramyotonic." Neurol. Centralbi.., 5:265-272, 1886.

Fontaine, J. et al.: "Hyperkalemic periodic paralysis and the adult muscle sodium channel alpha-submit gene." Science, 250:1000-1002, 1990.

George, A.L. et al.: "Assignment of a human skeletal muscle sodium channel alpha-submit gene (SCN4A) to 17q23.1-q 25.3." Genomics, 9:555-556, 1991.

Streeten, D.H.P.: Periodic paralysis. In: Stanbury, J.B., Wyngaarden, J.B., Frederickson, D.S. (Eds.): The Metabolic Basis of Inherited Disease. New York, McGraw-Hill, 1966.

AORTITIS SYNDROME WITH RHEUMATOID SPONDILITIS

Syndromography

Clinic

There are manifestations of aortic insufficiency in patients with rheumatoid spondylitis.

Paraclinical examinations

Echocardiography. To demonstrate valve lesions.

Blood. Research on rheumatoid arthritis.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Sokoloff, L.: The pathology of rheumatoid arthritis and allied disorders. In Hollander, J.L.(Ed.): Arthritis and allied conditions. Philadelphia, Lea & Febiger, 1966.

XIFHOID APPENDIX SYNDROME

Synonymy

S. of xiphoid hypersensitivity. S. of xiphoidalgia.

Syndromography

Clínic

There is deep pain in the chest, dull, of different intensity, from moderate to distressing, occasionally radiating to the epigastrium, back, shoulders, arm or precordium.

It occurs during the night and interferes with sleeping. Non-sudden installation, lasting from minutes to days. It recurs for weeks or months, rarely for years. Triggered by turning head, walking, getting up, stopping.

Physical examination. Palpation of the xiphoid process reproduces the typical pain and its associated manifestations.

Paraclinical examinations

Electrocardiogram. Normal

Stomach and duodenum radiology. Normal.

Cholecystography. Normal.

Syndromogenesis and etiology

It may appear associated with coronary artery disease, gastrointestinal diseases or only the gallbladder.

In cases where the xiphoid process has been removed, periostitis and perichondritis are reported.

The etiology is unknown.

Literature

Junghanns, H.: "Der Schwertfortsatzschemerz (Xyphoideodine)". Zentralbl. F. Chir., 67:628-629, 1940.

Lipkin, M.; THE. Fulton and E.A. Wolfson: "The syndrome of the hypersensitive xiphoid." New English J. Med., 253:591-597, 1955

CHONDROCOSTAL JOINT SYNDROME

Synonymy

S. of intercostal neuralgia.

Syndromography

Clinic

Pain referred to the costal or abdominal wall. Circulatory changes may occur in painful areas.

Syndromogenesis and etiology

It originates as a result of degenerative lesions, trauma or disease of the costovertebral joints with irritation of nerve roots or sympathetic ganglia.

The eleventh and twelfth dorsal nerves are frequently affected.

Literature

Goldthwaite, J.E.: "The rib joints." N. Engl. J. Med. 223:568-573, 1940.

ARTHROGRYPOSIS SYNDROME

Synonymy

S. multiple congenital arthrogryposis. S. of deforming fetal myodystrophy. S. de Stern Gueverin. S. of arthromyodysplasia. S. of Otto. S. de Rossi. S. de Rocher-Shetdon.

Syndromography

Clínic

It affects both sexes equally, limiting movement of all joints, except the mandibular and some spinal joints. Occasional paralysis of different muscles. Facial gesticulation is reduced and gives a look of sadness and melancholy.

Physical examination. Deformities of the joints with increased volume of the joints. Muscle hypoplasia and thickening of the neighboring skin (stiffness), alternating with atrophic areas.

Arms rotated inwards, elbows cylindrical. Fingers and wrists flexed. Legs rotated outward, hip dislocated. Knees extended. Premature synostosis of the skull and defects of the palate. Occasional absence of the sacrum.

Paraclinical examinations

Radiology. bone atrophies. The alterations referred to in the physical examination are checked. There are also: brachycephaly, jaw hypoplasia, osteoporosis and carpotarsal fusion after 10 years of age.

Muscle biopsy. Degenerative changes of muscles with fibrosis and fatty infiltration. Degeneration of motor neurons in the spinal cord.

Syndromogenesis and etiology

There are four pathogenic categories of arthrogryposis:

1. Myopathies.
2. Neuropathies.
3. Connective tissue diseases.
4. Exogenous defect: intrauterine space limitations or extrauterine pressures.

Three varieties linked to the X chromosome are distinguished, caused by different mutations and a different basic defect:

1. Lethal form with severe contractures, hypotonia, micrognathia and death due to respiratory failure (progressive loss of anterior horn cells).
2. Associated with ptosis, micropenis, cryptorchidism, inguinal hernia and normal intelligence (non-progressive intrauterine myopathy).
3. Arthrogryposis that improves with age, with normal intelligence and without other alterations (defect of the connective tissue of the tendons).

Literature

Beghin, J.: Les Aspects Radiologiques de L'arthrogrypose", J. Beige Radiology. 48:383, 1965.

Epstein, B.S.: "Radiogrhaphic identification of arthrogryposis multiplex congenita in utero." Radiology, 77:108, 1961.

Hall, J.G. et al.: "Three distinct types of X-linked arthrogryposis seen families", Clin. Genet., 21:81-97, 1982.

Hennekan, R.C.M. et al.: *A family with severe X-linked arthrogryposis". Eur. J. Pediat., 150:656-660, 1991.

Jacobson, H.G. et al.: "Arthrogryposis multiplex congenita". Radiology, 65:8, 1955.

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Ann. Pediatrician (Paris), 13:2-19, 1966.
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ARTHROPHTHALMOPATHY SYNDROME

Synonymy

S. of hereditary arthrophthalmopathy. S. of progressive arthrophthalmopathy.

Syndromography

Clinic

Arthrophthalmopathy syndrome occurs in childhood.

Progressive myopia appears, leading to blindness. Progressive reduction in hearing. Degenerative changes in several joints that can be severe.

Paraclinical examinations

Radiology. Large irregularities of the vertebral bodies of the thorax. Occasional scoliosis.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

David, B.: "Uber einen dominantn Erbgang bei enier polytopen enchondralen Dysostose Typ'. Pfaundler-Hurler. Ztschr. Orthop., 84:657-660, 1954.
Stickler, G.B. and D.G. Pugh: "Hereditatry progressive arthro-ophthalmopathy H. Additional observations on vertebral abnormalities, a hearing defect, and a report of a similar case."
Mayo Clin. Proc., 42:495-500, 1967.

BECKER SYNDROME

Synonymy

S. benign Duchenne muscular dystrophy. Duchenne late type.

Syndromography

Clinic

In this syndrome the symptoms are the same as those of Duchenne muscular dystrophy syndrome, but of late onset and slower progression.

Becker syndrome begins between 4-5 years of age. It begins with the pelvic belt and thighs (iliac muscles, psoas, glutes and quadriceps). It ascends progressively, reaching between 5 and 10 years the shoulder girdle and arms (pectorals, latissimus dorsi, trapezius, deltoids, biceps and finally sternocleidomastoid, spinal extensors and abdominals). There is early hypertrophy of the calves. Preserved intelligence.

Hypogenitalism sometimes.

Paraclinical examinations

Creatine phosphokinase. High in blood.

Electromyogram. Shows a typical pattern.

Muscle biopsy. There are fewer injuries and more regeneration.

ration.

Syndromogenesis and etiology

It is transmitted by sex-linked recessive inheritance (X chromosome). There is an alteration of the structure of the dystrophy unlike Duchenne muscular dystrophy in which the dystrophy is absent. The gene is located on Xp21.

Literature

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Cestan, R. et N. I. Leone:. Nouv. Iconogr. Salpetriere, 15:38, 1902.
Emery, A. and R. Skinner: "Clinical Studies in Benign. (Becker's Type X-linked muscular Dystrophy)". Clin. Genetics, 10:189, 1976.

BERTOLOTTI SYNDROME

Synonymy

S. of sacralization-scoliosis-sciatica.

Syndromography

Clínic

In Bertolotti syndrome there is numbness, hypersensitivity or pain along the sciatic, pain in the lower back, morning stiffness, scoliosis.

Paraclinical examinations

Radiology. Sacralization of the fifth lumbar vertebra (LV).

Syndromogenesis and etiology

It is produced by a sacralization of the LV vertebra.

Literature

Bertolotti, M.: "Contributo alla conoscenza dei vizi di differenzazione regionale del rachide con speciale riquardo all assimilazione sacrale della V lombare." Radiol. Med., 4:113-144, 1917.
Bertolotti, M.: "Le syndromes lombo-ischialgiques dórigine vertebrale." Rev. Neurol., 29:1112-1125, 1922.

BLOUNT-BARBER SYNDROME

Synonymy

S. of warm rod. S. of deforming osteochondrosis of the tibia. S. of medial or lateral osteochondrosis. S. de Erlacher-Blount.

Syndromography

Clínic

There are two forms of this syndrome: infantile and adolescent.

cent.

The infantile form occurs between the first and second year of age, generally in overweight children with gradual increase in bowing of the legs without apparent cause.

Almost always bilateral.

In the adolescent form, the same symptoms are observed as in the infantile form and they appear between 6 and 12 years of age.

Generally unilateral.

Physical examination. There is shortening of 1-2 cm of the affected leg or legs. Enlargement of the medial condyle. Internal rotation of the tibia.

Paraclinical examinations

Radiology. Irregularity of the contours of the proximal tibial epiphyseal line, areas of rarefaction in the metaphyses.

Syndromogenesis and etiology

There is a failure in the growth of the epiphyseal cartilage with delay in ossification of the lateral or medial portion of the proximal epiphysis of the tibia.

It has been suggested that Blount syndrome may be similar to chondrodysplasia punctata and that the changes in the tibia may be indistinguishable after the punctate calcification disappears.

The etiology is genetic, with autosomal recessive inheritance and multifactorial inheritance occurring in the childhood form.

Literature

Blount, W.P.: "Tibia vara, ostheochondrosis deformans tibiae." J. Bone Joint Surg., 19:1-29, 1937.

Ikegawa, S. et al.: Chondrodysplasia punctata mimicking Blount's disease: a case report." Acta Orthop. Scand., 61:580-581, 1990.

CAFFEY SYNDROME

Synonymy

S. of infantile cortical hyperostosis. S. of subperiosteal cortical hyperostosis. S. of infantile cortical hyperplasia. S. de Caffey- -Smith. S. by De Toni-Silverman-Caffey. S. de Roske-De Toni--Caffey.

Syndromography

Clinic

This syndrome is more common in women. It is established in early childhood (below 5 months), although occasionally bone lesions are detected, observed by ultrasound or radiographically, before birth. The lesions appear suddenly and are accompanied by irritability, fever, conjunctivitis, increased bone volume, discomfort and limitation of movement of the affected parts.

There is an increase in volume of the jaw and face, occasionally the increase in volume begins in the limbs and only after the face and other parts of the body are affected.

There are no changes in color, edema or increase in temperature in the skin surrounding the affected bones. There are no lymphadenopathy.

Paraclinical examinations

Radiology. Cortical thickening of the bone.

Blood. Moderate anemia, neutrophilia, accelerated erythrocyte sedimentation rate.

Urine. Normal.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is genetic. It has been suggested that vascular occlusion secondary to thrombocytosis could be involved in the pathogenesis.

Literature

Caffey, J. and W.A. Silverman: "Infantile cortical hyperostosis; preliminary report on a new syndrome". Amer. J. Roentgn., 54:1-16, 1945.

Caffey, J.: “Infantile cortical hyperostosis: a review of the clinical and radiographic features.” Proc. Roy. Soc. Med., 50:347- -354, 1956.

Pickering, D. and B. Cuddigan: "Infantile cortical hyperostosis associated with thrombocythaemia." Lancet, II:464-465, 1969.

CIRCUMSCRITAL CALCINOSIS SYNDROME

Synonymy

S. de Profichet.

Syndromography

Clinic

This syndrome is reported more frequently in adult women. There are primarily calcareous deposits on the limbs, symmetrical and most commonly affecting the periarticular tissue.

Paraclinical examinations

The same procedures as universal calcinosis should be performed.

Syndromogenesis and etiology

Many authors do not agree that this clinical form is an entity and consider that it is part of the universal calcinosis syndrome.

The etiology is unknown.

Literature

Leistyna, J.A. and A.H. Hassan: Interstitial calcinosis; report of a case and a review of the literature". Amer. J. Dis. Child., 107:96-101, 1964.

Reines, S.: "Petrificatio cutis circumscripta." Arch. Derm. Syph (Wein), 88:267-289, 1907.

UNIVERSAL CALCINOSIS SYNDROME

Synonymy

Fibrodysplasia ossificans progressiva of Teuts-

-Chiander. S. of diffuse calcinosis. S. of calcareous tendofascitis.

St. Pierre de la Peau"

Syndromography

Clinic

This syndrome affects both sexes, but is more common in females. It is established during the first two decades of life with variations depending on the extent of the abnormal calcium deposit in the skin, subcutaneous tissue, muscles, nerves, tendons, and organs. A vague history of non-specific fever is recorded.

There is weakness, pain and stiffness of the joints.

Physical examination. Irregular plaques are observed in the skin and deep tissues, most frequently located periarticularly, and only directly affecting the joints.

Initially there is no pain and the surrounding skin is normal. Later there is pain, and finally necrosis

and ulceration of the skin with discharge of whitish material and superinfection.

Paraclinical examinations

Radiology. Calcium deposits of various sizes and shapes, such as dense lines around muscles, tendons and nerves.

Blood. Normal.

Urine. Normal.

Syndromogenesis and etiology

Possibly due to polysaccharide disorders.

This syndrome has been reported associated with collagen diseases: scleroderma, dermatomyositis, lupus erythematosus, rheumatoid arthritis, myositis ossificans.

One third of cases are secondary to scleroderma or dermatomyositis.

The etiology is unknown.

Literature

Hilbish, T.E. et. al.: "Roentgen findings abnormal deposition of calcium in tissues". Am. J. Roentgenol, 87:1128, 1962.

Leistyna, J.A. and A.H. Hassan: “Interstitial calcinosis, report of a case and a review of the literature". Amer. J. Dis. Child. 107:96-101, 1964.

Massim, J.R. et al.: "Treatment of calcinosis universal with aluminum hydroxide." Arch. Dis. Child, 45: 118, 1970.

Moretti, E.: "Sur la calcinose generalisee." J. Radiol. Electrol. Med. Nuc., 45:433. 1964.

Teissier, LJ.: Du diabéte phosphatique. These pour le Doctorat du Médicine (Paris: Bailliere, 1876), p. 439.

CAMERA SYNDROME

Synonymy

S. of neuralgic osteopathy.

Syndromography

Clínica

This syndrome occurs in middle-aged patients.

It is characterized by pain that can affect practically any bone, the pain is intermittent and with progressive increase or sudden installation without apparent cause or after trauma. Difficult to specify its location due to the diffuse nature of the pain or its occasional irradiation. It is exacerbated at night. There is weight loss. There is no response to any medical, orthopedic or physical treatment.

Physical examination. There are painful points no larger than 1 cm on the affected bone, which can be detected with the tip of the finger and better located with a needle.

Paraclinical examinations

Radiology. Negative.

Syndromogenesis and etiology

There is localized fibrosis of the bone and hyperemia.

Literature

Bertola, L. and A. Pedrocca: "Oste opatie nevralgiformi lombosciatalgiche a localizzazioni vertebrali e paraver-tebrali (Camera Syndrome)". Minerva Orthop., 4: 215-218. 1953.

CAPLAN SYNDROME

Synonymy

S. of rheumatoid arthritis and pneumoconiosis. S. of silicoarthritis. S. de Collinet-Caplan.

Syndromography

Clinic

It occurs in patients generally exposed to fibrogenic dusts. There is migratory joint pain, edema, and other symptoms of rheumatoid arthritis that precede or follow lung pathology (cough, moderate dyspnea, hemoptysis).

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation. Latex and Waller Rose test positive.

Radiology. Typical joint disorders. Chest: shows 0.5-5 cm nodules, frequently peripheral, some with cavitation. Pulmonary fibrosis.

Syndromogenesis and etiology

These are patients with rheumatoid arthritis exposed to fibrogenic dusts. (Example: miners, quarry workers.)

Literature

Benedek, T.G.: *Rheumatoid-Pneumoconiosis". Am. J. Méd., 55: 515, 1973.

Capian, JA.: "Certain unusual radiological appearances in the chest of coal-miners suffering from rheumatoid arthritis" Thoray, 8: 29, 1953.

Caplan, A.; R.B. Payne and J.L. Withey: “A broader concept of Caplan's syndrome related to rheumatoid factors." Thorac., 17:205-212, 1962.

Christiaens, L. et al.: "Le Syndrome de Caplan-Collinet (a propos de 6 observations)". Arch. Mal. Prof., 15:546, 1954.

Edling, N.P. et al.: "Rheumatoid Pneumoconiosis (Caplan's disease). A report of three cases encountered in Sweden". Radio Act. (Diag.) (Stockh), 8:168, 1969.

Mattson, S.B.: "Caplan's syndrome in association with asbestosis", Scand. J. Resp. Dis.. 52:153, 1971.

Ramírez, R.J.; V. López-Majano and G. Schultze: "Caplan's syndrome, a clinicopathologie study." Amer. J. Med., 27:643- -652,1964.

CLUTTON SYNDROME

Synonymy

S. of Clutton joints.

Syndromography

Clinic

This syndrome generally presents insidiously between 6-16 years of age. The alterations are symmetrical and are characterized by chronic joint edema (85% in the knees) that is occasionally recurrent. In few cases it is acute or subacute in nature.

There is redness, pain, limitation of movement, fever and systemic manifestations, with polyarticular involvement.

Paraclinical examinations

Serology. Synovial fluid rich in lymphocytes.

Radiology. Negative.

Syndromogenesis and etiology

It is a congenital syphilis that produces chronic hydrarthrosis. There is no damage inside the joints.

Literature

Clutton, H.H.: “Symmetrical synovitis of the knee in hereditary syphilis.” Lancet, 1:391-393, 1886.
Hollander, J.L.: Arthritis and allied conditions, Philadelphia, Lea & Febiger, 1966.

COAL MINERS' ELBOW SYNDROME

Synonymy

S. from the students' elbow. S. of olecranon bursitis.

Syndromography

Clínica

Patients report pain in the elbow and a physical examination reveals discomfort, increased volume of the elbow and limitation of movement.

Paraclinical examinations

Radiology. Intra-articular calcifications.

Syndromogenesis and etiology

This syndrome is caused by repeated blows (jackhammer) or friction. It is associated with elbow diseases, gout, arthritis or diseases of unknown cause.

TENNIS ELBOW SYNDROME

Synonymy

S. of radiohumeral bursitis.

Syndromography

Clínica

Pain in the elbow, functional limitation, sensitivity to pressure in the radiohumeral area of the elbow joint.

Paraclinical examinations

They do not show alterations.

Syndromogenesis and etiology

The etiology is frequently associated with the practice of tennis. There is fatigue of the arm extensors.

Literature

Hollander, J.L.: Arthritis and allied conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.

COMPARTMENT SYNDROME

Compartment syndrome occurs when fluid accumulates at high pressure within a space closed by fascia (muscle compartment), decreasing capillary perfusion below the level necessary for tissue viability.

There are three main causes of compartment syndrome: increased fluid accumulation, decreased volume (compartment constriction), and limitation of volume expansion secondary to external compression.

Although compartment syndrome most commonly develops in all four compartments of the leg, it can also occur in the forearm, hand, arm, shoulder, foot, thigh, buttocks, and back. These are rarely diagnosed and that is why we are forced to make a short introduction for their better understanding, referring to the leg where they are most common.

The fascia of the thigh, fascia lata, continues into the leg where it is called the leg or crural fascia.

Deep extensions of this fascia form the septa:
anterior intermuscular, posterior intermuscular and transverse intermuscular. These septa and the interosseous membrane (tibia-fibula) make up the so-called compartments of the leg. They are: anterior compartment, lateral compartment, deep posterior compartment and superficial posterior compartment.

Each compartment houses certain muscles, arterial and venous branches, and nerves:

1 Anterior compartment: extensor muscles, tibialis anterior, extensor hallucis, extensor digitorum longus, and third peroneal muscles, which extend the toes and dorsally flex the foot; anterior tibial arteries and veins and deep peroneal nerve.
2. Lateral compartment: peroneal muscles and superficial peroneal nerve.
3. Deep posterior compartment: deep flexor muscles, flexor digitorum longus, tibialis posterior, flexor hallucis longus, popliteus, posterior tibial arteries and veins, peroneal artery and tibial nerve.
4. Superficial posterior compartment: superficial flexor muscles, soleus, gastrocnemius, arteries, veins and nerves.

It can now be better understood why different syndromes occur acutely or chronically in any of the compartments, in this case the legs.

ACUTE COMPARTMENT SYNDROME

Syndromography

Clinic

The most important symptom in an impending compartment syndrome is pain disproportionate to the initial injury. However, pain may be absent if there is also a disorder of the central or peripheral nervous system.

To remember the initial symptoms, it is useful to use the six P's rule of compartment syndrome:

1. Pressure. It is the earliest finding in a compartment that is swollen and tense on palpation.
2. Pain on extension (Pain). Passive movement of the fingers can cause pain in the ischemic muscles involved. The subjective aspect of the patient must be taken into account.
3. Paresis (Paresis). Muscle weakness may be a consequence of primary nerve involvement, muscle ischemia, or pain defense.
4. Paresthesia (Paresthesia). The most reliable physical finding in a conscious and cooperative patient is sensory deficit.

Initially, sensory detection manifests itself as

paresthesia, but if treatment is delayed, it can progress to hyperesthesia and anesthesia. Careful examination of sensitivity helps determine which compartments are involved.

5 and6. Pulses present (Pulse) and pink color (Pink). If there is no injury or severe arterial disease, peripheral pulses are palpable and capillary refill is generally preserved.

Paraclinical examinations

Needle technique.

Wick catheter technique.

Slit catheter technique.

All three allow the measurement of intracompartmental pressure that may be necessary in three groups of patients:

1. Patients who are uncooperative, untrustworthy, intoxicated by alcohol or drugs and in children.
2. Patients who are unresponsive due to head trauma or drug overdose.
3. Patients with associated neurovascular lesions, or neuropraxia due to associated nerve deficit.

Syndromogenesis and etiology

The most frequent causes of this syndrome are: fractures, vascular injury with intracompartmental hemorrhage, muscle swelling due to overexertion, exogenous fluid infiltration, and others.

Literature

Blick, S.S.; R.J. Brumback et al.: "Compartment syndrome in open tibial fractures." J. Bone Joint Surg., 68:1348-1353, 1986.

Matsem, FA.: Compartment Syndromes. Gune & Stratton. New York, 1980.

Mubarack, S.J.; AC Owen et al.: "Acute compartment syndromes: diagnosis and treatment with the aid of the wick catheter". J. Bone Joint Surg., 60A: 1091-1095, 1978.

ANTERIOR TIBIAL COMPARTMENT SYNDROME

Synonymy

S. of ischemic necrosis of the peroneal muscles. S. of the peroneal compartment.

Syndromography

Clinic

This syndrome can appear in one or both limbs and occurs after prolonged and continuous exercise. The patient reports fatigue in the legs, then edema and severe pain appear, which does not disappear with elevation of the legs or heat.

There is loss of dorsiflexion and inversion. Any movement produces pain.

A firm consistency is felt on palpation of the anterolateral portion of the leg, as well as absence of the pedis and posterior tibial pulses.

Anesthesia is observed in a triangular area on the dorsum of the foot, at the base of the first and second toes.

In chronic cases, recurrent pain appears after any effort and is only relieved with surgical decompression of the anterior compartment, through fasciotomy.

In acute cases, this emergency intervention must be performed to avoid irreversible muscle changes.

Paraclinical examinations

Radiology. To exclude trauma.

Syndromogenesis and etiology

It is caused by a decrease in blood flow to the muscles after severe exercise and is caused by ischemic swelling of the muscle within its tight fascial compartment.

Literature

Lunceford, E.M., Jr.: "The peroneal compartment syndrome." Southern Med. J., 58:621-623, 1965.

Reszel, P.A.; J.M. Janes and J.A. Spittel, Jr.: "Ischemic necrosis of the peroneal musculature: A lateral compartment syndrome; report of a case". Proc. Staff Meet. Mayo Clinic, 38:130-136, 1963.

Slocum, D.B.: The shin splint syndrome." Amer. J. Surg., 114:875-881, 1967.

METAPHYSEAL CHONDRODYSPLASIA SYNDROME (JANSEN)

Synonymy

S. of Jansen's metaphyseal dysostosis. S. of congenital metaphyseal dysostosis.

Syndromography

Clinic

Symptoms appear in childhood. There is marked dwarfism and physical retardation, hypertelorism and exophthalmia, in addition to contractures and deformities of the joints, particularly flexion of the hips and knees, as well as gradual edema of the joints and mental retardation.

Paraclinical examinations

Radiology. There is underdevelopment of the skull base, platybasia and brachycephaly, irregular mineralization of the mandible and the last ribs, clavicle and scapula, dwarfism and skeletal deformities in the adult.

Syndromogenesis and etiology

The etiology is genetic. There are many sporadic cases that are probably transmitted in an autosomal dominant manner.

nante.

CONRADI SYNDROME

Synonymy

S. of congenital calcifying chondrodystrophy. S. of epiphyseal punctate dysplasia. S. de Hünermann. S. with punctate epiphysis. S. of chondrodysplasia puntata type Conradi-Hünermann.

Syndromography

Clínica

This syndrome begins in the prenatal period and is only identifiable by radiology or ultrasound.

During childhood, visual disorders, delayed growth, congenital cataracts, contractures, shortening of the proximal long bones, thickened metaphyses, dwarfism, macrocephaly or microcephaly, syndactyly, mental deficiency, saddle nose root, micrognathia, kyphosis or scoliosis, dyskeratosis or ichthyosiform hyperkeratosis, seborrheic dermatosis occur.

There is a form called chondrodysplasia puntata rhizomelic type, which is very serious in which rhizomelia and psychomotor retardation occur. There is dwarfism, kyphoscoliosis, asymmetrical short limbs and contractures, facies with malar hypoplasia, epicanthus, general and external alopecia of the eyebrows, as well as of the scalp in some areas.

This variety is transmitted by autosomal recessive inheritance.

Paraclinical examinations

Radiology. Presence of multiple punctate calcium deposits in the epiphyses, carpus and tarsus, spine, sternum, posterior ribs. During childhood there are skeletal abnormalities.

Chromosome study. Normal.

Urine. Moderate aminoaciduria, nonspecific.

Syndromogenesis and etiology

It affects bones and ectodermal structures. It presents mucoid degeneration in spots, cystic spaces in the epiphyses and calcifications. Likewise in the lenses.

There are several syndromes with similar bone alterations:

recessive and dominant chondrodysplasia punctata linked to the

Literature

Comings, D.E.: C. Papazian and H.R. Schoene: "Conradi disease:

"chondrodystrophia calcificans congenita: congenital stippled epiphyses". J. Pediat., 72: 63, 1968.

Conradi, E.: "Vorzeitiges. Auftreten Von Knochen und eigenartigen verka kung kermen Bei chondrodystro fhia foetalis hypoplastica. Histoligische." Und Roentg even ter Suchungen.

Jahb. Kinderheilk. 80: 86, 1914.

Gwinn, J.L. et al.: "Conradi disease: (chondrodystrophia calcificans congenita." Am J. Dis. Child., 121: 503, 1971.

Josephson Bonyoriati, M.D.: "Chondrodystrophia calcificans congenita. Report of a case. Review of the literature." Pediatrics, 28: 425, 1961.

Wardinsky, T.D. et al.: "Rhizomelic chondrodysplasia punctata and survival beyond one year: a review of the literature and five case reports." Clin. Genet., 38:84-93, 1990.

DUPUYTREN CONTRACTURE SYNDROME

Synonymy

S. of contraction of the palmar fascia.

Clunic

Syndromography

This syndrome is more common in men. It has a high family incidence and a gradual increase with age, especially after age 40. It can be unilateral or bilateral.

It frequently affects the right hand. The fingers are affected in the following order: small, middle and index. There is pain in the palm of the hands and progressive loss of function due to the inability to extend the fingers.

Syndromogenesis and etiology

It is transmitted by inheritance. It has been observed in several generations of the same family and is associated with many pathologies such as epilepsy, pulmonary tuberculosis, and chronic alcoholism.

The etiology is genetic. The basic defect has not yet been specified.

Literature

Dupuytren, G.: "Permanent retraction of the fingers, produced by an affection of the palmar fascia." Lancet, 2:222, 1833-1834.

Hollander, J.L.: Arthritis and allied conditions. Philadelphia, Lea & Febiger, 1966.

Young, ILD. andR.W. Fortt: "Familial fibromatosis." Clin. Genet., 20:211-216, 1981.

CAST CORSET SYNDROME

Syndromography

Clinic

This syndrome is characterized by prolonged nausea and repeated vomiting. It occurs in patients who, as indicated by the orthopedic doctor, use a plaster corset on their body.

Paraclinical examinations

Blood. Hypokalemic alkalosis, hypovolemia.

Syndromogenesis and etiology

It is considered to be due to mechanical compression of the fourth portion of the duodenum by the superior mesenteric artery, resulting in gastric and duodenal dilation.

Literature

Dorph, M.H.: "The cast syndrome: Review of literature and report of a case." N. Engl. J. Med., 243:440-442, 1950.

Nelson, J.P.; DO. Ferris and J.C. Ivins: "The cast syndrome, case report." Postgrad. Med., 42:457-461, 1967.

CERVICAL RIB SYNDROME

Synonymy

S. of the first supernumerary rib.

Syndromography

Clinic

This syndrome almost always appears in adulthood, after shoulder trauma due to stretching or straining the arm, or during pregnancy. It is generally unilateral although supernumerary ribs can be bilateral and more frequently in women. There is pain in the ulnar border that increases with movements, weakness of the hands, arm, shoulders, numbness and loss of sensation.

Physical examination. There is evidence of: cyanosis, edema, coldness, vasomotor spasms, muscle atrophy, weak pulse on the affected side, occasionally a bone mass is palpable in the supraclavicular fossa on that

Paraclinical examinations

Radiology. Shows the cervical supernumerary rib, it must be differentiated from the cervical disc.

Syndromogenesis and etiology

It is produced by the supernumerary cervical rib, which irritates the brachial plexus, compressing it between the rib and the anterior scalenus muscle.

Due to the familial nature, genetic etiology with autosomal dominant inheritance has been proposed.

Literature

Holst, S.: "Cervical rib and associated vascular complications." J. Oslo City Hosp., 13:173-182, 1963.

Lishman, W.A. and W.R. Russell: "The brachial neuropathies." Lancet, 2:941-946, 1961.

Murphy, T.: “Brachial neuritis caused by pressure of first rib." Aust. Med. J., 15:582-585, 1910.

Schapera, J.: “Autosomal dominant inheritance of cervical ribs.” Clin. Genet., 31:386-388, 1987.

Smyth, C.J.: "Rheumatism and arthritis." Ann. Int. Med., 59 suppl. 4, 74-75, 1963.

Willshire, J.: "Supernumerary first rib." Lancet, 2:633, 1860.

TRANSIENT COXITIS SYNDROME

Synonymy

S. of observation hip. S. of acute transient epiphysitis. S. of transient synovitis. S. of coxitis fugax.

Syndromography

Clinic

It occurs in children, there is limping accompanied or not by referred pain to the knee, thigh, and groin.

There is slight limitation of passive movements.

Paraclinical examinations

Radiology. Normal.

Blood. Normal erythrocyte sedimentation rate.

Node biopsy. Slight hyperplasia.

Syndromogenesis and etiology

This syndrome is induced by minor trauma, focal infections, allergies. More frequently it is idiopathic.

It usually occurs due to osteoarthritis or synovitis.

Literature

Hunder, G.G. and P.J. Kelly: Roentgenologic transition osteoporosis of the hip: a clinical syndrome." Ann. Int. Med., 68:539-552, 1968.

Lovett, R.W. and J.L. Morse: “A transient or ephimeral form of hip disease with report of cases." Boston Med. & Surg. J., 127:161-163, 1892.

CROUZON SYNDROME

Synonymy

S. of craniofacial dysostosis. S. of craniostenosis. S. of Virchow occicephaly. S. of occicephaly-acrocephaly. Parrot-headed S.

Syndromography

Clínica

This syndrome occurs more frequently in males, more often during the first year of birth.

Patients report headache, moderate hearing loss, and progressive vision loss.

Physical examination. Skull: brachycephaly, scaphocephaly or trigonocephaly. Forehead wide, flat and with frontal prominences. Sometimes torricephaly.

Facies: hypoplasia of the middle facial mass, short upper lip and protrusion of the lower lip, oblique palbebral cleft, effaced lower orbital ridges, exophthalmos, divergent strabismus, papilledema, hypertelorism, low-set ears, narrow duct orifice.

Ogival palatine vault, partial anodontia.

Sometimes moderate mental retardation due to intracranial hypertension. Seizures may occur.

Paraclinical examinations

Radiology. Coronary and sagittal craniosynostosis. Cranial base short, sloping. Widened Turkish chair. Deep cerebellar fossa. Small maxillary sinuses.

Cerebrospinal fluid. Increase in pressure.

Syndromogenesis and etiology

There is premature closure of the cranial sutures and secondarily brain injury due to intracranial hypertension.

The etiology is genetic: a third of cases are sporadic, due to a fresh mutation. The majority is transmitted by autosomal dominant inheritance with variable expressivity and reduced penetrance.

There is a recessive form.

Literature

Crouzon, O.: "Dysostose cranéofaciale hereditaire." Bull. Mem. Soc. Med. Hóp (Paris) 33:545, 1912.

Dodge. H.W. Jr.; N.W. Wood and R.L.J. Kenedy: "Craniofacial dysostosis; Crouzon Disease." Pediatrics 23:98, 1959.

Dube, B.K.; K.C. Singhal; B. Kishore et al.: "Crouzon's syndrome." Indian Pract.. 20:402-408, 1967.

Flippen, J.H. Jr.: "Craniofacial dysostosis of Crouzon." Pediatric, 5: 90, 1950.

Rubinstein, J.H.: In Gellis-Kagaw. Current Pediatric Therapy. 7th ed. Saunder, Philadelphia, 1976.p. 409.

CYRIAX SYNDROME

Synonymy

S. de Colleg-Davies. S. of the sliding rib.

Syndromography

Clinic

The patient reports chest pain that may be dull and recurrent or very sharp and associated with symptoms of shock. It can occur after a sneeze, a deep breath, or a movement of the arms.

Compression of the cartilage of the affected rib reproduces the symptoms.

Syndromogenesis and etiology

The involvement of the interchondral synovial membrane of the three false ribs (8th, 9th and 1st), results in the end

The anterior rib remains under the cartilage and compresses the intercostal nerve and sympathetic fibers.

The etiology may be traumatic or idiopathic.

Literature

Camishion, R.C. and F. Goldstein: “Partial noncalculous cystic duct obstruction (cystic duct syndrome)". Surg. Clin. N. Amer., 47:1107-11 14, 1967.

Schieden, V.: "Uber die 'Stauungsgallenblase" Zentralbl. Chir., 41:1257, 1920.

SYNDROME OF INTERNAL DISARREMENT OF THE KNEE JOINT

Syndromography

Clínica

Pain on knee movements, localized discomfort, thigh muscle atrophy, joint edema.

Paraclinical examinations

Radiology.

Joint puncture and fluid study.

Synovial membrane biopsy.

Syndromogenesis and etiology

It is a syndrome that can be produced by different causes:

Meniscus cysts.

Calcifications.

Tibial plate fracture.

Fracture of the kneecap.

Traumatic synovial effusion.

Chondromalacia of the patella.

Literature

Wilson, P.D.: In Hollander, J.L., ed.: Arthritis and allied conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.

CRANIODIPHYSEAL DYSOSTOSIS

Syndromography

Clínica

This syndrome affects both sexes, starting in childhood, then progresses to the symptomatic form within the first years. It is accompanied by nasal obstruction, anorexia, vomiting, blindness, deafness and mental retardation.

Physical examination. You can also see facial distortion, widened ribs, and thickened clavicles in their middle part.

Paraclinical examinations

Blood. Normal calcium, phosphorus and alkaline phosphatase.

Bone radiology.

Syndromogenesis and etiology

Of genetic etiology, it is considered to be transmitted by autosomal recessive inheritance. Autosomal dominant inheritance is also proposed.

The basic defect is unknown.

Literature

Gorlin, R.J. and H. Sedano: "Craniometaphyseal dysostosis and craniodiaphyseal dysostosis." Mod. Med., 36:154-155, 1968.

Halliday, J.: “A rare case of bone dystrophy." Br. J. Surg., 37:52- -63, 1949.

PERIPHERAL DYSOSTOSIS SYNDROME

Synonymy

S. of epiphysometaphysal acrodysplasia (acrodysostosis).

Syndromography

Clínica

There is shortening of the width of the hands and feet. Slight to moderate growth retardation.

Paraclinical examinations

Radiology. The conical shaped metacarpals are observed.

Premature closure of the growth cartilage that produces deformation of the fingers and toes.

Syndromogenesis and etiology

Of genetic etiology and autosomal dominant inheritance.

The basic defect is unknown.

Literature

Brailsford, J.E.: The radiology of bones and joints. London-Churchill, 1948.

Cooper, R.R. et al.: “Metaphyseal dysostosis: description of an ultrastructural defect in the epiphyseal plate chondrocites.” J. Bone Joint. Surg: 55:485, 1973.

Gledion, A.: Acrodysplasia, in Kaufmann, H.J.: Progress in pediatric radiology. v. 4, Basel-Karger, 1973.

Graces, L. et al.: *Peripheral dysostosis: investigation of metabolic and endocrine functions". J. Pediatric, 74: 730, 1969.

Hernández, R.M.; A. Miranda and S. Kofman-Alfaro: *Acrodysostosis in two generations: an autosomal dominant syndrome". Clin. Genet. 39:376-382, 1991.

Singleton, E.B. et al.: “Peripheral dysostosis". Am.J. Roentgenol., 84:499, 1960.

HIP-SHOULDER DYSPLASIA SYNDROME

Synonymy

S. of Becken-Schuiter dysplasia.

Syndromography

Clinic

In this syndrome, the following occur in a variable, not constant, way: microphthalmia, retinal coloboma, and coneal opacities.

Paraclinical examinations

Radiology. Hypoplasia of the scapula, iliacus, clavicle and lordosis of the lumbosacral spine. Failure in rib development.

Syndromogenesis and etiology

It is probably transmitted by autosomal dominant inheritance. te, and it is possible that it is a new mutation.

The etiology is unknown.

Literature

Kosenow, W.; J. Mederce and A. Sinios: "Becken-Schuiter dysplasie", Fortsch. Röntgenstr, 113-139, 1970.

MULTIPLE EPYPHYSARY DYSPLASIA SYNDROME

Synonymy

S. of polyepiphyseal dysplasia. S. of polyepiphyseal osteochondral dystrophy. Poliosteochondritis. S. of Fairbank.

Syndromography

Clinic

Clinical manifestations often present in late childhood or adolescence, then the diagnosis can be made. There is limping, pain and stiffness in the hips, knees and ankles and limited movement of the joints.

Frequently there are coxa vara and genus valgum or varus.

Patients have a height below 152 cm.

Paraclinical examinations

Radiology. Delay in the appearance of the secondary ossification centers of the long bones and the centers of the flat bones of the hands and wrists.

The epiphyses of the long bones can be small, irregular and in some cases flattened; these lesions are more pronounced in the lower limbs.

The skull, sternum, ribs and collarbones are generally not affected.

Syndromogenesis and etiology

There is a chondrodystrophy of the cartilaginous epiphyses. There are multiple ossification centers developing at different periods.

This syndrome is transmitted by autosomal dominant inheritance.

The etiology is genetic.

Literature

Barrington-Ward, L.E.: Double cOxa vara with other deformities occurring in brother and sister". Lancet 1: 157, 1912.

Caffey, J.: “Achondroplasia of pelvis and lumbosacral spine.” Am. J. Roentgenol., 80:449-457, 1958.

Fairbank, H.A.T.: "Generalized disease of skeleton." Proc. R. Medical Soc. 28:1611, 1935.

Jacobs, P.: Multiple epiphyseal dysplasia in Kaufmann. H.J. Progress in Pediatric Radiology, vol. 4 Basel-Karger 1973, p. 309.

Murphy, M.C. et al.: "Multiple epiphyseal dysplasia." J. Bone Joint Sug. 55A:814, 1973.

Shapira, E.; E. Fischel; S. Moses et al.: "Syndrome of incomplete regional achondroplasia (ilium and ribs) with abdominal muscle dysplasia." Arch. Dis. Child., 40:694-697, 1965.

HEMIMELIC EPYPHYSAL DYSPLASIA SİNDROME

Synonymy

S. of osteochondroma of the epiphysis. Trevor's disease.

Fairbank disease.

Syndromography

Clinic

There is unilateral and asymmetric hard edema of the knee or ankle with or without pain, or restriction of motility. Varus or valgus according to the affected site.

Other locations: epiphysis of the femoral head, long bones and wrist.

Paraclinical examinations

Radiology. Overgrowth on one side of the epiphysis, with an irregular contour and distal ossification of the femur, tibia and heel; also the proximal portion of the tibia.

Syndromogenesis and etiology

It is more common in the male sex. It does not appear to have a Mendelian inheritance pattern.

The etiology is unknown.

Literature

Donaldson, J.S. et. al.: Osteochondroma of the distal femoral epiphysis". J. Pediatr. 43, 212, 1953.

Fairbank, T.J.: "Dysplasia epiphysealis hemimelica." J. Bone Joint. Surgery. 38B:237, 1956.

Mouchet, A. et al.: "La Tarsomegalie." J. Radiol. Electrol. 10:289, 1926.

Osman, M.Z. et al.: "Dysplasia apiphysealis hemimelica". Semin. Roentgenol., 8:174, 1973.

KOZLOWSKI SPONDYLOEPYPHISAL DYSPLASIA SYNDROME

Synonymy

S. of spondylometaphysal dysostosis.

Syndromography

Clinic

The alterations are observed in the preschool stage. There is moderate dwarfism, more marked on the trunk. Short neck. Scoliosis or kyphoscoliosis. Shortening of hands and feet. Normal-poor craniofacial appearance. Limitation of joint movements with gait disorders.

Paraclinical examinations

Radiology. Widening, sclerosis and irregularities in the metaphyses of long bones. Shortening of the iliac bones. Delay in skeletal maturation. Kyphosis and kyphoscoliosis.

Syndromogenesis and etiology

There are some cases of dominant hereditary transmission. It has been suggested that the genetic defect affects collagen (COL 2A1).

The etiology is genetic.

Literature

Kozlowski, K; P. Maroteaux and J. Spranger: "La dysostose Sponaylo-metaphisaire". Presse Méd. 75: 2769. 1967

Koz, K.: Spondylo-metaphyseal dysplasia in Kaufmann, H. J. Progress in Pediatrics. Radiology, vol. 4 Basel-Karger, 1973, p. 299.

Piffareti, P.G., et al.: "La dysostose spondylo-metaphysaire de Kozlowski, Maroteaux et Spranger." Ann. Radiol. (Paris) 13:405, 1970.

Riggs, W. Jr. et al.: "Spondylometaphyseal dysplasia: Report of affected mother and son." Radiology, 101: 375, 1971.

Tiller, GE., et al.: "Tandem duplication within a type II collagen gene (COL 2A1) exon in an individual with spondylo-epiphiseal dysplasia." Proc. Nat. Acad. Sci., 87:3889-3893, 1990.

BONE DYSPLASIA-RETINAL DETACHMENT-DEAFNESS SYNDROME

Syndromography

Clinic

Bilateral retinal detachment during the first year of life and blindness. Hearing loss between 6-12 years old. Hearing loss for high frequencies increases.

Paraclinical examinations

Radiology. Loss of the trabeculae of the metaphyses and epiphyses of long bones, which produces a shortening of them in coxa vara. Deformity of the femoral head, kyphoscoliosis. Vertebral crushes and pelvic distortion.

Laboratory exams. All negative.

Syndromogenesis and etiology

It is a syndrome of genetic etiology that is transmitted by inheritance linked to the X chromosome, and in which the clinical manifestations are mild in heterozygous women.

Literature

Roaf, R.; J.B. Longmore and R.M. Forrester: "A childhood syndrome of bone dysplasia, retinal detachment, and deafness." Develop. Med. Child. Neurol., 9:464-473, 1967.

DISPLASE SYNDROME PSEUDOACHONDROPLASTIC

Synonymy

S. of spondyloepiphyseal dysplasia, pseudoacondoplastic type.

Syndromography

Clínic

In this syndrome, patients present short-limb dwarfism, generally detected in the second to fourth years of life. Marked shortening of hands and feet. Normal facial appearance.

Paraclinical examinations

Radiology. Normal craniofacial bones. Shortening of long bones. Fragmentation and irregularities of the developing epiphyses. Biconcave vertebral bodies. Coxa vara. The ribs have a spatula appearance.

Literature

Cooper, R. R. et al.: "Pseudoachondroplastic dwarfism. J. Bone. Joint Surg. 55:475, 1973.

Ford, N. et al.: "Spondylo-epiphyseal dysplasia." Am.J. Roentgenol. 86;462. 1961

Lindseth, R.E. et al.: "Spondylo-epiphyseal dysplasia." Am. J. Dis. Child, 113-721, 1967.

Maroteau, P. and M. Lamy: "Les formes pseudo-achondroplastique des dysplasies spondylo-epiphysaires." Presse Méd. 67: 383, Silverman, 1959.

F.N.: "Pseudoachondroplastic dysplasia." Semin. Roentgenol., 8: 172, 1973.

MUSCULAR DYSTROPHY SYNDROME DE LIMB-GIROLE

Synonymy

S. Erb's juvenile muscular dystrophy. S. of Moebius Leyden muscular dystrophy.

Syndromography

Clínica

Symptoms appear at any age but generally around the age of 20. There is seizure of the proximal muscles of the arms in the ERB type, more rarely in the lumbosacral region (Leyden-Moebius type).

Paraclinical examinations

Blood. Enzymes: G.O. transaminase, aldolase and creatine phosphokinase show a variable elevation: in the initial stage there is a significant elevation. Later it is normal or there is slight elevation.

Electroencephalogram. Normal.

Syndromogenesis and etiology

It can be transmitted by autosomal recessive and autosomal dominant inheritance. It has been suggested that in these cases there are also proteindystrophin alterations.

Literature

Arikawa, E. et al.: "The frequency of patients with dystrophim abnormalities in a limb-girdle patient population." Neurology, 41:1491-1496, 1991.

REFLEX SYMPATHETIC DYSTROPHY SYNDROME

Syndromography

Clínica

It is seen more frequently in patients over 50 years of age. Weeks or months after a triggering factor such as local trauma, heart attack, peripheral nerve injuries or stroke, the first symptoms appear on palpation in the limbs, which may be:

spontaneous, burning pain and swelling, accompanied by signs and symptoms of vasomotor instability and atrophic changes in the skin.

The limb is hot, edematous and hypersensitive, particularly in the periarticular region. Afterwards the skin becomes shiny and cold and flexion contractures of the hand or foot appear.

The shoulder on the affected side often becomes painful with limited movement (shoulder-hand syndrome).

It can affect the kneecap and hip.

Paraclinical examinations

Radiology. There is demineralization of bones. The radiograph of the affected distal part shows osteopenia in the form of a "spot" (Sudeck's atrophy).

Radionuclide bone scan. It shows greater uptake in the periarticular bone of the affected side.

Syndromogenesis and etiology

This syndrome occurs in older patients and is preceded by some associated triggering factor already mentioned.

There is involvement of sympathetic nerves.

Literature

Kosin, F. et al.: "Tite reflex sympathetic dystrophy syndrome (RSDS), Am. J. Med.. 70: 23, 1982.

LANGE TYPE I MUSCULAR DYSTROPHY SYNDROME

Synonymy

S. of Amsterdam type dwarfism. S. type Amstelo-damensis.

S. Lange type. S. Brachman-De Lange.

Syndromography

Clinic

Physical examination. In patients affected by this syndrome, hirsutism of the face, back and forehead is observed. There is brachycephaly or microcephaly, long eyebrows, generally alternating exotropia, low set of ears, small nose, mouth with separated teeth, ape-like hands, small, curved, somewhat rudimentary or absent fingers with differences between the two hands. Feet with partial or total syndactyly. There is mental retardation.

Limited vocabulary or does not speak, walks with help or alone. Weight and development markedly affected.

Paraclinical examinations

Study of the chromosome pattern.

Blood. Normal.

Urine and cerebrospinal fluid. Normal.

Nitrogen balance and basal metabolism. Generally normal.

Syndromogenesis and etiology

It is a group of congenital anomalies without a genetic basis. In some cases chromosomal abnormalities have been reported.

The etiology is unknown.

Literature

De Lange, C.: “Sur un type nouveau de dégenération (typus Amstelodamensis)”. Arch. Med. Enfant., 36:713-719, 1933.

Falek, A.; R. Schmidt; G.A. Jervis: "Familial De Lange syndrome with chromosome abnormalities." Pediatrics, 37:92-101, 1966.

Hart, Z.H.; RI. Jaslow and M.R. Gómez: "The de Lange syndrome." Am. J. Dis. Child., 109:325-332, 1965.

ANTERIOR ABDOMINAL WALL PAIN SYNDROME

Syndromography

Clinic

There are two subtypes according to the location of the pain: right or left lower quadrant and upper region of the upper quadrant.

The pain is continuous, sometimes related to movement, and is not related to ingestion of food or defecation.

Physical examination. With the patient lying supine, the painful area is firmly compressed with a single finger or thumb, and the patient is asked to elevate both legs a few inches. If the pain increases due to muscle contraction, it must be pain originating in the wall. If it decreases it must be visceral.

Syndromogenesis and etiology

It may be associated with other disorders, but is independent of gastrointestinal disorders.

The etiology is unknown.

Literature

Long, C.: "Myofacial pain syndromes." Henry Ford Hosp. Med. Bull., 4:102-106, 1956.

PAIN SYNDROME IN THE ANTERIOR TIBLAL REGION

Synonymy

S. de Maroch.

Syndromography

Clinic

Pain in the muscles of the anterior tibial region even at rest. Numbness and paresthesias in the foot are common.

There is loss of sensation in the feet and legs, redness, edema and tenderness in the anterior tibial region. Paralysis may be a late symptom.

Physical examination. Induration and swelling of the anterior compartment are the earliest signs. There is a decrease in capillary refill of the fingers and paleness of the foot. Lately, absence of pulsations and neurological deficit may appear.

Paraclinical examinations

Muscle biopsy. Various degrees of necrosis.

Measurement of pressure in the compartment. Generally- high.

Syndromogenesis and etiology

Etiology: muscle damage or compression of the tibial artery and lymphatics. Anterior tibial nerve injury? Spasm, thrombosis or embolism of the anterior tibial artery? accumulation of fluid in the anterior tibial compartment.

It is currently considered among the compartment syndromes caused by increased pressure inside a closed compartment of the limb. The anterior compartment of the leg is most frequently affected.

Literature

Bhild, C.G., III: “Noinfective gangrene following fractures of lower leg." Ann. Surg., 116:721-728, 1942.

Paton, D.E: "The pathogenesis of anterior tibial syndrome." J. Bone Joint Surg., 50B:383-385, 1968.

BROAD DORSAL SYNDROME

Syndromography

Clinic

The patient reports pain in the lower back and lower limbs. Occasionally associated with pain in the upper back, shoulders, upper limbs, neck and chest.

Injection with local anesthesia of the thoracolumbar fascia suppresses pain (this has diagnostic value).

Syndromogenesis and etiology

There is irritation of the sensory fibers of the aponeurosis of the latissimus dorsi muscle due to fibrosis of the subfascial fat, as well as adhesion of the tissue to the fascia and defect of the latter.

Literature

Copeman, W.S.C. and W.L. Ackerman: "Edema or herniations of fat lobules as a cause of lumbar and gluteal fibrositis." Arch. Int. Med. 79:22-35, 1947.

Dittrich, R.J.: The latissimus dorsi syndrome". 0hio Med. J.. 51:973-975, 1955.

DUPLAY SYNDROME

Synonymy

Adhesive capsulitis of the shoulder. S. of scapulohumeral periarthritis. S. of periarticular fibrosis of the shoulder. S. of adhesive bursitis. S. of periarticular fibrositis.

Syndromography

Clínica

It is more common in women over 40 years of age, rarely before menopause. It occurs in elderly men and sedentary workers. There is a gradual increase in pain in the shoulder during abduction and internal rotation. The pain radiates to the arm and forearm, occasionally to the scapula.

Physical examination. Scapulohumeral fixation. Arm in adduction. Pain on palpation in various areas of the shoulder.

Paraclinical examinations

Radiology. Negative.

Two or three months after the syndrome sets in, there is demineralization of the humerus.

Syndromogenesis and etiology

Etiology: 85% is idiopathic. 15% post-traumatic.

Frequently observed after a period of inactivity and muscle spasm, in anxiety and depression.

This syndrome is associated with visceral diseases.

Literature

Duplay, E.S.: "De La peri-arthrite scapulo-humerale et des raideurs de lépaule qui en sont la consequence." Arch. Gen. Med., 6,20:513-542, 1872.

Hollander, J.L.: Arthritis and allied conditions. Philadelphia, Lea & Febiger, 1966.

ENGELMANN SYNDROME

Synonymy

S. of progressive diaphyseal dysplasia. S. de Camurati-Engelmann. S. of infantile multiple hyperostotic osteopathy.

Syndromography

Clinic

It affects both sexes. Manifestations are more frequent in the first 6 years of life, more rarely in older children and early adolescence. There is delay in walking, difficulties in gaining weight, anorexia, weakness in the legs.

Physical examination. Poor development, elongated legs, muscle atrophy, symmetrical bilateral fusiform lengthening of the diaphysis of the long bones, genus varus. genus valgus, coxa valga.

It less frequently affects the short bones, ribs and pelvis. It does not affect bones of the face.

Paraclinical examinations

Blood. Normal calcium, phosphorus and alkaline phosphatase.

Radiology. Cortical thickening, fusiform osteosclerotic enlargement of the diaphysis of the bones. Normal epiphysis.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is genetic, but the basic defect is unknown.

Literature

Camurati, M.: "I gave a rare case of osteite simmetrica ereditaria degli arti inferiori." Chir. Org. Movim., 6:662-665, 1922.

Engelmann, G.: “Ein von osteopathia hyperostotica (sclerotisans), multiplex infantilis.” Fortschr. Geb. Rontgehstr., 39:1101- -1 106, 1929.

Joseph, R.; J. Lefevbre; E. Guy, et al.: "Dysplasia craniodiaphysaire progressive." Ann. de Rad., 1:477-490, 1958.

Sparkes, R.S. and C.B. Graham: "Camurati-Engelmann disease. Genetics and clinical manifestations with a review of the literature". Am. J. Med. Genet., 9:73-85, 1972.

SCAPULOCOSTAL SYNDROME

Syndromography

Clínic

This syndrome occurs in middle-aged patients, is insidious in nature and is accompanied by deep pain in the shoulder, with different irradiations: neck, occipital region, chest or combinations of these.

Physical examination. Signs of tenderness appear from the reference point or below the superior medial angle of the scapula to the posterior chest wall.

Paraclinical examinations

Shoulder and cervical spine x-rays.

Syndromogenesis and etiology

The etiology may be of traumatic origin or produced by arthritis, bursitis or myositis.

Literature

Michele, A.A.: "Scapulocostal syndrome its mechanism and diagnosis." New York J. Med. 55:2485-2493, 1955.
Hollander, J.L.: Arthritis and allied conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.

LIGAMENTOUS OSSIFICANS SPONDILITIS I SYNDROME

Synonymy

S. of physiological vertebral ligamentous calcification.

Syndromnography

Clinic

It is observed in patients over 50 years of age with moderate or absent back symptoms, but with decreased mobility of the spine. There is no evidence suggestive of rheumatoid arthritis.

Paraclinical examinations

Bone x-ray. One or multiple affected intervertebral spaces are seen. The thoracic region is the one that most frequently presents calcifications of the ligaments that take on a tortuous appearance. However, normal vertebrae show normal density, as well as normality in the vertebral and costovertebral processes and joints.

Erythrosedimentation. Normal.

Syndromogenesis and etiology

In this syndrome there are degenerative changes followed by calcification of the longitudinal vertebral ligaments, which lead to limitations in motility.

Literature

Oppenheimer, A.: "Calcification and ossification of vertebral ligaments (spondylitis ossificans ligamentosa) roentgen study of pathogenesis and clinical significance." Radiology, 38:160- -173, 1942.

Smith, C.E; D.G. Pugh and H.E. Polley: "Physiologic vertebral ligamentous calcification: an aging process." Am. J. Roentgenol., 74:1049-1058, 1955.

MULTIPLE EXOSTOSIS SYNDROME

Synonymy

S. of Ehrenfried. S. of diaphysal aclasis. S. of multiple cartilaginous exostoses.

Syndromography

Clinic

There is deformity of the limbs, including the hands (short pasterns) and short stature. It also affects ribs and scapulae, never the skull. There are nerve and spinal cord compressions as a complication.

Prominent hard tumors are seen on the knees, scapulae, costal arches, shoulders, wrists, and ankles. There is shortening of the limbs involved, bulging of the arms and legs, there may be a discrepancy in the length of the limbs. Shortening of height, in the male adult 169 cm and in the female 159 cm. Although these tumors may be present at birth, they are generally not appreciated until later, growing slowly until adolescence and stopping in adulthood. There is an incidence of sarcoma of 2-10% in adults.

Paraclinical examinations

Radiology (bone survey). Juxtaepiphyseal diaphyseal cartilaginous exostoses are seen in long bones, fundamentally, ulna, radius, humerus, tibia and fibula, involving joints, pelvis, scapulae and ribs.

Blood. Normal.

Syndromogenesis and etiology

It is due to a diaphysal-juxtaepiphysial overgrowth covered by hyaline cartilage.

It is transmitted by autosomal dominant inheritance. Twenty to 50% of patients develop malignant changes, although it has recently been estimated that malignancy only occurs in 0.5 to 2% of cases.

The etiology is genetic but the basic defect is unknown.

Literature

Ehrenfried, A.: "Multiple cartilaginous exotoses hereditary deforming chondrodysplasia. A brief report on a little known disease." J.A.M.A., 64:1606-1642, 1915.

Hennekem, R.C.M.: “Hereditary multiple exostoses.” J. Med. Genet., 28:262-266, 1991.

Jaffe, H.L.: “Hereditary multiple exostoses.” Arch. Path.. 36:335-357, 1943.

Solomon, L.: "Hereditary multiple exostoses." Am. J. Hum. Genet., 16: 351-353, 1964.

BONE FIBROGENESIS IMPERFECTA SYNDROME

Syndromography

Clinic

The patient reports progressive bone pain that sets in in adulthood. There is weakness, muscle atrophy, contractures.

Paraclinical examinations

Radiology. There is pseudofracture of ribs, cortical thinning, and additional degenerative changes in joints.

Blood. Increased serum alkaline phosphatase.

Literature

Baker, S.L. and H.M. Turnbull,: Two cases of hitherto undescribed disease characterized by a gross defect in the collagen of the bone matrix", J. Pathol. Bacteriol. 62:132, 1950.

Baker, S.L. et al.: "Fibrogenesis imperfecta ossium." J. Bone Joint Surgery. 48-B, 804, 1966.

Frame, B.: C.Y. Fost: W. Pack: W. Reynolds and R.J. Argen: "Fibrogenesis imperfecta ossium: a collagen defect causing osteomalacia". N. Engl. J. Med. 285:769, 1971.

FRONTODIGITAL SYNDROME

Syndromography

Clínic

There is widening of the big toe and fingers in most cases. Syndactyly and/or polydactyly in some.

Paraclinical examinations

Radiology. Craniosynostosis is missing. There is widening of the distal phalanges of the big toes and alterations of the other toes.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance with variable expression.

The etiology is genetic but the basic defect is unknown.

Literature

Marlham, R.E. and D.W. Smith: "Frontodigital syndrome. A dominantly inherited disorder with normal intelligence." J. Pediatrician 77:129, 1970.

KNEE BLOW SYNDROME

Synonymy

S. for housemaid. S. of the coal miners. S. of prepatellar bursitis.

Syndromography

Clínic

There are three clinical forms of this syndrome:

1. Acute form: there is local pain, edema and local tenderness.
2. Chronic form: there is a chronic distention of one of the prepatellar bursae, usually the one above the tibial tubercle or tuberosity.
3. Infected form: it is due to a secondary infection of a pre-existing bursitis, with all the signs of infection.
   The most common causes are a wound located over the patellar region or an abrasion.

Paraclinical examinations

Knee x-ray.

Syndromogenesis and etiology

The causes, already indicated, are: trauma, unaccustomed work on the knee, a long period kneeling, direct or indirect infection of the bursa.

Literature

Smillie, L.S.: Injuries of the knee joint. ed. 4, Baltimore, Williams & Wilkins, 1962.

HALLERMANN-STREIFF SYNDROME

Synonymy

S. of mandibulo-oculofacial dyscephaly. S. of dyscephaly. S. by Ullrich. S. of Fremerey-Dohna.

Syndromography

Clinic

It affects both sexes equally. It is characterized by localized alopecia, prominent nose, microconea, congenital cataracts with spontaneous rupture and absorption, occasionally glaucoma.
There is harmonic dwarfism, skin atrophy. Delayed psychomotor development.

Paraclinical examinations

Radiology. Reveals the features in the jaw. There is ramus hypoplasia, anterior displacement of the temporomandibular joint. The condyle may be completely absent.
The skull is brachycephalic. Delayed closure of the fontanels, small face, small orbits.

Syndromogenesis and etiology

It is possibly transmitted by autosomal recessive inheritance, although it appears that all reported cases are sporadic.

Viral infection has been raised during pregnancy or alterations caused by teratogenic substances. Genetic etiology is suggested.

Literature

Cohen, M.M. Jr.: "Hallermann-Streiff syndrome: a review." Am. J. Med. Genet., 41:488-499. 1991.
Hallermann, W.: "Vogelgesicht und Catarcta congenita." Klin. Monatsbl. Augenh., 113:315-318, 1948.
Kulrander, G.J.; N.W. Lavy and J. A. Campbell: "Roentgen differentiation of the oculodentadigital syndrome and the Hallermann-Streiff syndrome in infancy." Radiology, 86:77-86, 1966.
Streiff, E.B.: "Dysmorphie mandibulo faciale (tete d'oiseau) et alterations oculaires." Ophthalmologica, 120:79-83, 1950.

HENCH-ROSENBERG SYNDROME

Synonymy

S. of palindromic rheumatism. S. de Hench.

Syndromography

Clinic

It is characterized by a sudden attack of moderate to severe pain, followed a few hours later by an increase in joint volume. Attacks occur at any time of the day, but most often in the afternoon and return completely within a day or two.

Physical examination. There is involvement of the fingers, knees, hips, shoulders, ankles, temporomandibular joint, sternoclavicular joint and, less frequently, the cervical spine.

Occasionally there is periarticular or soft tissue inflammation. Sudden appearance in some patients of nodules that disappear within a week.

Paraclinical examinations

Blood. Moderate lymphocytosis and elevated erythrocyte sedimentation rate during the attack. All tests for rheumatoid arthritis are negative. Occasional increase in lipids and fatty acids.

Literature

Hench, P.S. and E.F. Rosenberg: *Palindromic rheumatism; new off recurring disease of joints (arthritis, periarthritis, para-arthritis) apparently producing no articular residues-report of 34 cases; its relationship to angioneural arthrosis, allergic rheumatism and rheumatoid arthritis". Arch. Int. Med. 73:293-321. 1944.

Lamond-Havres, R.W.: Palindromic rheumatism. In Hollander, J.L.: Arthritis and allied conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.

HYPERTROPHY SYNDROME

OF MASSETERS

Synonymy

S. of benign masseteric hypertrophy.

Syndromography

Clínic

There is unilateral or bilateral hypertrophy of the masseter muscles accompanied by painful edema below and in front of the ear with or without pain or temporomandibular arthralgia. Teeth grinding during sleep.

Patients generally have a tense personality.

Paraclinical examinations

Radiology. There is mandibular distortion when the hypertrophy is unilateral. Dental malocclusion.

Syndromogenesis and etiology

Acquired condition produced by different mechanisms:

Primary or secondary malocclusion, displacement of the jaw due to movements during reading, tension and teeth grinding.

Literature

Barton, R.T.: "Benign masseteric hypertrophy; a syndrome of importance in the differential diagnosis of parotid tumors." J.A.M.A., 164:1646-1647, 1957.

Gurney, GE.: "Chronic bilateral benign hypertrophy of masseter muscles." Am. J. Surg., 73:137-139, 1947.

HOFFA SYNDROME

Synonymy

S. hypertrophy of the infrapatellar fat pad.

Syndromography

Clínic

There is usually a long history of pain in the knee compartment related to exercise.

Physical examination. There is edema due to the presence of increased fat pad, moderate effusion. Upon forced extension, pain and local sensitivity to deep pressure occur.

Paraclinical examinations

Knee radiology. It allows us to rule out bone injury and in many cases an increase in soft and intra-articular tissues can be observed.

Syndromogenesis and etiology

There is hypertrophy of the fat cushion that causes alterations of the synovial bursa between the condyles during foot extension.

Literature

Hoffa, A.: "The influence of adipose tissue with regard to the pathology of knee joint." J.A.M.A., 43:795-796, 1904.

JAFFE SYNDROME

Synonymy

S. of pigmented villonodular synovitis. S. polymorphocellular benign synovial membrane tumor. Giant cell tumor of the tendon sheath.

Syndromography

Clínic

There is pain in one or several large joints, with functional limitation of the joint. The knee is affected more frequently.

Physical examination. There is moderate limitation of the movements of the joints affected by the pain.

Paraclinical examinations

Joint radiology.

Synovial membrane biopsy.

Blood. Tests to rule out rheumatoid arthritis.

Erythrosedimentation.

Syndromogenesis and etiology

It has been suggested that its presence is secondary to neoplasms and traumas.

The etiology is unknown.

Literature

Chung, S.M.K. and J.M. Jones: Diffuse pigmented villonodular synovitis of hip joint." J. Bone Surg., 47A:293-303. 1965.

Jaffe. H.L.: L. Lichtenstein and C.J. Sutro: Pigmented villonodular synovitis, and bursitis, tenosynovitis. "A discussion of the synovial and bursal equivalents of the tenosynovial lesion commonly denoted as xanthoma, xanthogranuloma, giant-cell tumor, or myeloplaxoma of the tendon, sheath, with some consideration of the tendon sheath lesion itself."

Arch. Path., 31:731-765, 1941.

JAFFE-LICHTENSTEIN SYNDROME

Synonymy

S. of monostotic fibrous dysplasia.

Syndromography

Clínica

In healthy children, an increase in volume, discomfort and pain appears in a long bone, facial bone or skull.

No other skeletal manifestations appear.

Paraclinical examinations

Radiology. There are cystic areas in the cortex of the bones.

Blood. Occasionally, a slight elevation of calcium and alkaline phosphatase is observed.

Syndromogenesis and etiology

It may be a congenital anomaly or possibly a disruption of the normal reparative process after trauma.

The etiology is unknown.

Literature

Jaffe, H.L. and L.Lichtenstein: “Non-osteogenic fibroma of bone". Am. J. Path., 18:205-215, 1942.

Ross, D.W. and C.C. Vitale: “Monostotic fibrous dysplasia of the metacarpal." J. Bone Joint Surg., 37A:196-200, 1955.

JANSEN SYNDROME

Synonymy

Jansen type metaphyseal dysplasia.

Syndromography

Clínica

This syndrome manifests itself in both sexes. Dwarfism is evident since childhood with involvement of the metaphysis of all injured bones with marked widening. The shafts are shortened.

Paraclinical examinations

Radiology. The abnormalities already described are observed. The condyles appear irregular. The cuneiform vertebrae, with irregular contours.

Syndromogenesis and etiology

It has not been determined whether or not it is genetic.

The etiology is unknown.

Literature

Burkhart, J.M: E.C. Burke and P.J. Kelly: “Birth defects.” Mayo Clinic Proc., 40: 481-489, 1965.

Jansen, M.: "Uber atypische Condrodystrophie (Achondroplasie) und uber eine noch nicht beschriebene angeborene Waschstumtoring des Knochensystems: Metaphysare Dysostosis." Zeitschr. F. Orthop. Chir., 61:253-286, 1934.

KASHIN-BECK SYNDROME

Synonymy

Urov's disease. Tokut-Ze disease.

Syndromography

Clínica

It is an example of regional pathology. It occurs in childhood. It is endemic in Korea, China, Siberia. Asymptomatic or painful installation. There is muscle weakness, cramps, paresthesias, fatigue.

From 6 months to a year, joint stiffness occurs. There is a classification in degrees according to the joint intake and clinical manifestations:

First degree: moderate form, in the initial stage.

Second degree: increased joint pain, pain and general symptoms.

Third degree: chronic deforming arthritis and general manifestations.

There is weight loss, seizure of the spine and chronic gastritis. There are also deformities of the wrist and interphalangeal joints, crepitation. There is no inflammation or effusion. It is symmetrical in location and slow progression of osteoarthritis to other joints. There is muscle atrophy adjacent to the joint socket.

Paraclinical examinations

Radiology. It shows a dystrophic process of tubular growth of bones and shortening of the limbs.

Blood. Normal. In the third degree there is anemia and lymphocytosis. Normal calcium and phosphorus.

Syndromogenesis and etiology

Of toxic origin secondary to the ingestion of cereal grains infected with the fungus Sporotrichiela fusarium.

Literature

Beck, E.B.: "To the problem of disforming endemic osteoarthritis in the Baikal area." Russian Physician, 5:74-75, 1906.

Kashin, N.I.: The description of the endemic and other diseases, prevailing in the Urov-river area. The records of physicomedical scientific society attached to the Moscow Univ., Jan. 3, 1859.

Nesterov, A.I.: "The clinical course of Kashin-Beck disease." Arthr. Rheumat., 7:29-40, 1964.

KILOH-NEVIN SYNDROME

Synonymy

S. of the anterior interosseous nerve.

Syndromography

Clinic

Spontaneous installation or after trauma.

There is isolated paralysis of the flexor pollicis longus and the flexor digitorum profundus. There is no sensitive commitment.

Syndromogenesis and etiology

In terms of etiology, it is considered that it may be idiopathic or associated with fractures or damage to the bones of the forearm.

Bibliography

Farber, J.S. and R.S. Bryan: The anterior interosseous nerve syndrome." J. Bone Joint Surg., 50:521-523, 1969.

Kiloh, L.G.; S. Nevin: “Isolated neuritis of anterior interosseous nerve.” Br. Med. J., 1: 850-85 1, 1952.

Parsonage, M.J. and J.W.A. Turner: "Neurologic amyopathy. "The shoulder-girdle syndrome". Lancet, 1:973-978, 1948.

KLIPPEL-FEIL SYNDROME

Synonymy

S. of congenital bone torticollis. S. of congenital synostosis of the cervicothoracic vertebrae. S. of congenital fusion of the cervical vertebrae.

Syndromography

Clinic

The fusion of one or more cervical vertebrae consists of a congenital malformation that sometimes occurs asymptomatically. When the fusion is multiple, they shorten the cervical spine and give a characteristic appearance of a short neck with a low head sitting on the shoulders with limited movement and a low hairline, often at the level of the first thoracic vertebra. It is frequently associated with other abnormalities such as lack of scapular descent (Sprengel's deformity), platybasia, basilar impression, Arnold-Chiari deformity, etc.

"Mirror movements" (in which voluntary movements of one upper limb are accompanied by similar movements in the opposite limb) have been observed.

There is dyspnea and dysphagia. This syndrome is occasionally associated with neurological disorders due to congenital dysplasia of the spinal cord.

Physical examination. There is shortening of the neck. Platybasia and limited neck movements. It is occasionally associated with torticollis. There is facial asymmetry, scoliosis and kyphosis.

It can be subdivided into three types:

Type 1. Extensive anomalies with elements from different vertebrae integrating a simple block.

Type 2. Lack of complete segmentation of one or two cervical intervertebral spaces.

Type 3. Includes types 1 and 2, coexisting abnormalities of the lower thoracic and lumbar spine.

Paraclinical examinations

Radiology. It demonstrates vertebral shortening and fusion, as well as associated malformations.

Syndromogenesis and etiology

Four types of vertical fusion are currently recognized.
brocervical:

Type I. Massive fusion of cervical and upper thoracic vertebrae en bloc.

Type II. Fusion of only one or two vertebrae (possible hemivertebrae, atlantoccipital fusion and other anomalies).

Type II. Cervical, lower thoracic and lumbar fusion.

Type IV. Cervical fusion and other vertebrae and agenesis of the sacrum.

In type II, the subtype with C2-C3 fusion has autosomal dominant inheritance and the C5-Có subtype shows an autosomal recessive pattern.

This clinical heterogeneity can be based on genetic heterogeneity, however in general sense it is considered as an entity of genetic etiology with inheritance.

autosomal dominant with reduced penetrance and variable expressivity.

Literature

Daas-Rothscild, A. et al.: "Klippel-Feil anomaly with sacral agenesis: an additional subtype, type IV". J. Craniofac. Genet. Div. Biol., 8:297-301, 1988.

Gardner, W.J.: "Diastematomyelia and the Klippel-Feil syndrome", Clevel Clin. Quat.. 31:19-44, 1964.

Gunderson, C.H.; RH. Greenspan; G.H. Glaser et al.: "The Klippel-Feil syndrome: genetic and clinical reevaluation of cervical fusion." Medicine, 46:491-512, 1967.

KNIEST SYNDROME

Synonymy

S. of bone dysplasia-retinal impairment-deafness.

Syndromography

Clinic

There is out-of-proportion dwarfism, round face, prominent eyes, cleft palate, myopia, deterioration of the retina, cataracts, blindness, progressive conduction deafness, limitation of joint motility, pain in the large joints, kyphoscoliosis, lordosis, short neck, abnormal gait.

Paraclinical examinations

Radiology. Shortening of long bones. Loss of normal trabecular pattern. Glass appearance of bones.

Syndromogenesis and etiology

It is a syndrome of genetic etiology that is transmitted in an autosomal dominant pattern.

When examined with the electron microscope, the cartilage presents abnormalities in the organization of the collagen fibers. These are much thicker than normal and irregular in shape, without the characteristic banded pattern.

The loss of protopeptide C, which is required for the normal formation of type II collagen fibers, has been suggested.

Literature

Poole, A.R., et al.: "Kniest dysplasia is characterized by an apparent abnormal processing of the C-propeptide of type II cartilage collagen resulting in imperfect fibroid assembly." J. Clin. Invest., 81:579-589, 1988.

KOCKER-DEBRÉ-SEMELAIGNE SYNDROME

Synonymy

S. de Debré-Semelaigne. S. of cretinism. S. of hypertrophy muscular.

Syndromography

Clinic

It occurs in children and adults. There are typical manifestations of cretinism associated with a feeling of stiffness and discomfort in the large muscles, some movements are painful.

roses There is slowness muscle contraction. The walk is rough and slow. Cold temperature contributes to slowing of movements (paramyotonia). They present dysarthria secondary to the enlargement of the tongue. There is muscle hypertrophy.

Paraclinical examinations

Electromyogram. Pathological.

Urine. There is no creatinuria.

Blood. High cholesterol.

Iodine uptake. Decreased as well as other functions thyroid.

Syndromogenesis and etiology

Family incidence has been reported.

The etiology is unknown.

Literature

Debré, R.; G. Semelaigne: "Syndrome of diffuse muscular hypertrophy in infants causing athletic appearance. Its connection with congenital myxedema." Amer. J. Dis. Child., 50:1351-1361, 1935.

Hoffman, J.: "Weiterer Beitrag zur Lehre von der Tetanie, Deutsch. Ztschr. Nervenh. 9:278-290, 1896.

Kocker, T.: ""Zur Verhuntung des Cretinismus und cretinoider Zustande nach neuen Forschungen." Deutsch. Z. Chir., 34:556- -626, 1892.

LANDOUZY-DÉJERINE SYNDROME

Synonymy

S. of fascioscapulohumeral dystrophy. S. of scapulohumeral dystrophy.

Syndromography

Clínica

It is installed between 12-14 years old, until 30-40 years old.

Both sexes are affected equally. The first symptoms are: weakness in the movement of the shoulder muscles. Women present it when combing their hair. Difficulty closing eyes and blowing. Later, the muscles of the pelvis and legs are affected. There are winged scapulae.

Paraclinical examinations

Electromyogram. Pathological.

Urine. There is slight creatinuria.

Blood. Decreased creatinine.

Syndromogenesis and etiology

The etiology is genetic and is transmitted by autosomal dominant inheritance linked to chromosome 4 in q35 and it is estimated that it has a penetrance of 95% at the age of 20 years.

Biochemical analyzes detected a reduction in electron transport activity through complex II-IV.

There is no evidence of genetic heterogeneity.

Literature

Landouzy, L. and Déjerine, J.: "De la myopathie atrophique progressive; myopathie héréditaire, sans neuropathie, debutant d'ordinaire dans l' enfance par la face." Paris, F. Alcan., 1885.

Lunt, P.W. and P.S. Harper: "Genetic counseling in fascioscapulohumeral muscular dystrophy." J. Med. Genet., 28:655-664, 1991.

Mathews, K.D., et al.: "Characterization of the fascios- capulohumeral dystrophy locus on 4q35 (Abstract)." Am.J. Med. Genet. 49(suppl.) 350-350, 1991.

Shy, GM.: Neuromuscular disorders. In: Beeson, PM., McDermott, W., ed.: Cecil Loeb Textbook of Medicien, ed. 12, Philadelphia, Saunders, 1967.

Slipetz, D.M., J.R. Aprille: P.R. Goodyer and R. Rozen: "Deficiency of complex III of the mitochondrial respiratory chain in a patient with fascioscapulohumeral disease." Am. J. Med. Genet., 48:502-510, 1991.

LEGG-CALVÉ-PERTHES SYNDROME

Synonymy

S. of osteochondrosis of the capital femoral epiphysis. S. of flat coxa. S. de Calvé-Perthes. S. de Legg-Perthes.

Syndromography

Clinic

The symptoms are sudden or progressive, and appear between 6-12 years of age, with moderate pain in the hip, limitation of movement and leg lameness that progresses progressively and the pain becomes more intense.

Physical examination. Discomfort and muscle spasm of the hip. Possible atrophy of muscles and shortening of the legs.

Paraclinical examinations

Blood. Slightly accelerated erythrocyte sedimentation rate.

Radiology. Initially there is bone resorption and then sclerosis. Fragmentation of epiphysis. Irregular joints.

Syndromogenesis and etiology

There is idiopathic ischemia of the ossification centers. It can be traumatic.

The etiology is unknown.

Literature

Legg, A.T.: "The cause of atrophy in joint disease." Am.J. Orthop. Surg., 6:84-90, 1908-1909.

LERI SYNDROME

Synonymy

S. of pleonosteosis.

Syndromography

Clinic

It affects both sexes. It settles in early childhood. Mental development is generally normal. Some cases with altered intelligence have been reported. There is limitation of movements of joints and spine. Carpal tunnel syndrome appears. Morton's metatarsalgia syndrome may appear.

Physical examination. There is mongoloid facies (inconstant), widening and deformity of the index fingers and thumb, short and thickened hands, flexion contracture of interphalangeal joints, semiflexion and internal rotation of upper limbs, semiflexion and external rotation of lower limbs.

Paraclinical examinations

Radiologist. The bone alterations that cause the indicated deformities are checked.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is genetic, but the basic defect is Know.

Literature

Leri, A.: "Une Dysstrophie osseuse généralisée et hereditaire la plenasteose familiale." Press. Med., 30:13-16, 1922.

LEWIS-BESANT SYNDROME

Synonymy

S. of localized congenital muscular dystrophy.

Syndromography

Clinic

Cases have been reported in girls, as well as boys who died at an early age with respiratory failure.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Lewis, A.J. and D.E. Besant: “Muscular dystrophy in childhood.” J. Pediatr., 60:376-384, 1962.
Zellweger, H. A. Afifi; WE McCormick and W. Mergner: "Benign congenital muscular dystrophy." Clin. Ped., 6:655-663, 1967.

MAFFUCCI SYNDROME

Synonymy

S. of dyschondroplasia with hemangioma. S. of chondrodystrophy with hamartoma. S. vascular. S. of Kast.

Syndromography

Clinic

Patients are asymptomatic at birth. During childhood, bone and joint deformities appear.

It affects both sexes. There is no history of pain. There is orthostatic hypotension in sitting or standing positions. Fractures occur with minimal trauma.

Physical examination. Before puberty, nodules appear on the toes, later tumors occur in the cylindrical bones, with a symmetrical distribution. Hemangiomas appear in the subcutaneous tissue and soft tissue. There is no visceral commitment.

Paraclinical examinations

Radiology. Bone survey.

Arteriography and venography.

Skin and bone biopsy.

Syndromogenesis and etiology

There is a combination of hemangiomatosis and enchondromatosis and it does not appear to be hereditary.

The etiology is unknown.

Literature

Elmore, S.M. and W.C. Cantrell: "Maffucci's syndrome." J. Bone Joint Surg., 48A: 1607-1613, 1966.
Maffucci, A.: "I gave a case of enchondroma and multiple angoima. "Contribution to the embryonic genes of the tumor." Movimento Medico-Chirurgico, 3:399-412, 1881.

SHOULDER MUSCULOTENDINE CUFF SYNDROME

Syndromography

Clinic

There is usually a prior history of trauma to the shoulder.

The patient reports feeling a burst, accompanied by sudden pain, frequently followed by an asymptomatic and subsequently recurrent period that results in the impossibility of raising the arm or initiating abduction.

There may be chronic symptoms, given by redness of the shoulder, functional impotence for motility and discomfort at night when turning to that side.

Physical examination. Localized changes in skin color, shoulder pain, and crepitus may be observed.

Paraclinical examinations

Radiographic examinations. They are negative.

Syndromogenesis and etiology

A tear of the musculoskeletal cuff of the shoulder occurs with periarticular soft tissue injuries caused by trauma.

Literature

Hollander, J.L.: Arthritis and allied conditions. 7 ed., Philadelphia, Lea & Febiger, 1966.

STERNAL MANUBRIUM SYNDROME

Syndromography

Clínica

It can occur in patients with rheumatoid arthritis or without changes in the joints.

The patient reports pain above or on each side of the sternal manubrium. The pain is produced or aggravated by the effort of walking, climbing stairs, coughing, etc.

Physical examination. Tenderness and occasionally edema of the sternal manubrium joint that is relieved with procaine infiltration.

Paraclinical examinations

Radiology. Occasionally there are slight changes in the handlebar joint. Generally there are no changes.

Electrocardiogram. Normal.

Syndromogenesis and etiology

This syndrome occurs as part of rheumatoid arthritis syndrome or in isolation.

The etiology is unknown.

Literature

Fisher, C.M. and W. Light: "Manubriosternal arthalgia." N. Engl. J. Med., 256:799-801, 1957.

Sodestrom, N.: "Manubrial pain and angina pectoris." Svensk. Lakartidn., 48:1845-1847, 1951.

MARIE-BANBERGER SYNDROME

Synonymy

S. de Banberger. S. de Hagner. S. of secondary hypertrophic osteoarthropathy. S. of hypertrophic pulmonary osteoarthropathy.

Syndromography

Clínic

It is characterized by the presence of newly formed periosteal bone, increased joint volume with enlargement of the epiphyses of long bones, digital clubbing and arthritis. Deformity of the nails that present cyanosis. There is a sensation of heat in the tips of the fingers. Sweating of hands and feet. Arthralgia. The distal phalanges are widened and clubbed.

Digital clubbing can appear in primary or secondary forms. The primary appears in the familial form and the secondary is often accompanied by chronic bronchitis, diffuse lung diseases and bacterial endocarditis.

Familial osteoarthropathy known as pachydermoperiostosis is characterized by thickening of the skin of the face, scalp, and limbs.

Paraclinical examinations

Study of synovial fluid. There are less than 500 cells/mm² with a predominance of mononuclear cells.

Radiology. It shows periosteal thickening with new bone formation. Distal phalanges with hypertrophic changes and osteolysis in advanced stages.

Radionuclide study with pericortical linear uptake.

Syndromogenesis and etiology

In adults, the syndrome is generally secondary to some lung neoplasm, but rarely occurs in chronic lung or liver diseases (cirrhosis), cardiac, gastrointestinal, chronic infectious or neoplastic diseases.

It can occur idiopathic and familial.

The cause is unknown; Correction of the syndrome by vagotomy suggests vagal stimulation.

It has also been proposed that it may be related to circulating vasodilators, circulating immune complexes and hormones.

ute

The etiology is unknown.

Literature

Von Banberger, E.: "Ueber Knochenveranderungen bei chronischen Lungen und Herzhrankheiten." Zschr. Klin. Med., 18:193-217, 1891.

Fischer, D.S.; D.H. Singer and S.M. Feldman: "Clubbing, a review with emphasis on hereditary acropachy." Medicine, 43:459-479, 1964.

Marie, P.: "De lósteoarthropathie hypertrophiante pneumique." Rev. de Méd., 10:1-36, 1890.

MARFAN SYNDROME

Synonymy

S. of congenital mesodermal dystrophy. S. of dolichostenomelia. S. of arachnodactyly.

Syndromography

This syndrome is due to an alteration of the connective tissue and is characterized by great clinical variability. It affects three fundamental systems: skeletal, ocular and cardiovascular:

however, intelligence is not affected.

The diagnosis is made based on the most obvious manifestations: subluxation of the lens, dilation of the ascending aorta, dissecting aneurysm and ductal ectasia. The diagnostic criteria, according to an international committee of consultants from 1986, are:

1. If there is a first-degree relative affected by the syndrome, the patient under study must have: a) at least two systems involved; b) present a greater manifestation (this requirement will be age-dependent and according to the peculiarities of the family phenotype).
2. If there is no first-degree relative unequivocally affected by the syndrome, the patient under study must have: a) involved the skeletal system; b) involved at least two of the other systems; c) at least one major manifestation.

Clinic

The body of these patients is thin and elongated, the skull dolichocephalic, the ears prominent. They have a pointed palate, elongated arms and legs, hands with long and thin fingers, kyphoscoliosis, excavated chest, flat feet and hammer toes. The adipose panniculus is decreased. There is hyperextensibility and joint dislocation, muscle hypotonia. Ocular alterations are: myopia, strabismus, miosis, nystagmus, lens subluxation, coloboma. There are cardiovascular alterations such as: valvular deformities, septal defect, aneurysm of the aorta and pulmonary arteries. There are also lung and kidney defects.

Paraclinical examinations

Urine. Nitroprusside test to rule out homocystinuria.

Eye examination with slit lamp and full dilation. To detect small subluxations.

Echocardiography. Measure especially aortic root.

Computerized axial tomography. For the diagnosis of the dural sac at the level of L5, which appears in two thirds of affected patients.

Radiological examinations (bone survey). Large perinasal sinuses, dolichocephaly, elongated facial bones, excessively long and thin long bones, high vertebrae, etc.

Blood. Decrease in mucoproteins.

Other tests: karyotype, hemoglobin electrophoresis. To rule out other causes of tall stature.

Syndromogenesis and etiology

It is a syndrome of genetic etiology and is transmitted by autosomal dominant inheritance. The gene has been located on chromosome 15 around q21 and is due to mutations in the fibrillin gene (FBN1), a glycoprotein of extracellular microfibrils, which are associated with elastin. This alteration explains the basic defect of the syndrome.

Literature

Eldridge, R.: "The metacarpal index: a useful aid in the diagnosis of the Marfan syndrome." Arch. Int. Med., 113:248-254, 1964.
Grahame, R. and R.E. Pyeritz: The Marfan syndrome: joint and skin manifestations are prevalent and correlated." Br. J. Rheumatol., 34:126-2 13, i995.
Marfan, A.B.: "Un cas de déformation congenitale des four members, plus prononcée aux extrémités, caracteriséempar lállongement des os avec un certain degre dámincissement." Bull. Medical Soc. Hop. de Paris, 13:220-226, 1896.
Pyeritz, R.E.: The Marfan syndrome. In Royce, P.M. and B Steinman.: Connective tissue and its heritable disorders. Wily-Liss, 1933, pp 437-468.

DIABETIC MYELOPATHY SYNDROME

Synonymy

S. of diabetic amyotrophy.

Syndromography

Clinic

It affects both sexes in diabetic patients in the fifth to seventh decades. Pain occurs in the legs: severe, asymmetrical, occasionally unilateral, and preferably located in the hip and thigh.

Physical examination. Asymmetrical wasting of the muscles, fasciculations and absence of tendon reflectivity. There is loss of sensation and ulcers. Normal paleness in the ankles. Presence of Babinski sign.

Paraclinical examinations

Lumbar puncture. Increased protein.

Electromyography. Partial denervation of the leg muscles. There is no fibrillation.

Syndromogenesis and etiology

This syndrome is primarily motor in contrast to diabetic pseudotabes which is sensory.

The etiology is diabetes mellitus.

Literature

Bruns, L.: “Ueber neuritische Lahmungen beim Diabetes Mellitus.” Berl. Klin. Wschr., 27;509-515, 1890.
Garland, H. and D. Taverner: "Diabetic myelopathy." Br. Med. J. 1:1405-1408, 1953.
Grinker, R.R. and A.L. Sahs: Neurology, ed. 6, Springfield, Thomas, 1966.

IDIOPATHIC PAROXYSTIC MYOGLOBINURIA SYNDROME

Synonymy

S. of primitive myoglobinuria. S. de Meyer-Betz.

Syndromography

Clinic

It predominates in the male sex (4:1). It is installed at any age. In 50% of cases it appears after exercise, in the rest the cause is unknown.

The patient suddenly reports severe pain and muscle cramps with subsequent appearance of weakness and paralysis, accompanied by muscle swelling.

They generally affect the muscles of the legs and thighs, rarely the arms, speech, swallowing or breathing.
These crises can last a few hours. The general condition is affected and may lead to disorders of consciousness. One to three hours after the crisis, the elimination of clear urine is seen, but of a bloody pink or brown color, which gives a positive reaction to benzidine. The intensity of the urine color increases in the following hours and then fades.

During severe attacks, fever, skin and mucous hemorrhages, intense headache, asthenia, vomiting, paleness and abdominal pain may appear. On some occasions during the paroxysm of the crisis, acute renal failure occurs, with oliguria, isosthenuria, and edema that can lead to myoglobinuric renal shock. Mortality due to kidney failure is around 20%.

Physical examination. Muscle atrophy may occur after repeated attacks. Previously, the muscles appear edematous, painful and of a woody consistency.

Paraclinical examinations

Urine. A few hours after the attack, the urine is initially pinkish in color and then brown-red.
Oliguria and anuria in some cases. There is presence of myoglobin, cylindruria, erythrocyturia and leukocyturia.

Positive reaction to benzidine.

Muscle biopsy. Performed after the attack, it shows coagulation necrosis and discolored necrotic fibers alternating with normal fibers.

Blood. Increased muscle enzymes in serum.

Syndromogenesis and etiology

The alterations are located in muscle cells that present a marked sensitivity to trauma or unknown metabolic products. Histologically there are signs of degeneration in the muscle cells.

Cases have been reported in families.

The etiology is unknown.

Literature

Meyer-Betz, F.: "Beobachtunger an einem eigenartigen, mit Muskellähmunger Vesbunden Fall von Hämoglobinurie." Arch. Klin. Med. 101:86-127, 1911.
Rainey, R.L.; PW. Estes; C.L. Neely, et al.: "Myoglobinuria following diabetic acidosis." Intern Arch. Med., 111:564-571, 1963.

HYPERTROPHIC MYOPATHY SYNDROME

Synonymy

S. of hypertrophy musculorum vera. S. of true muscle hypertrophy.

Syndromography

Clinic

Predominates in men. It usually begins in childhood, adolescence or early adulthood, vague onset with painful sensations in the affected muscles.

Physical examination. Slow growth of muscles in the limbs that are affected more frequently than the trunk. However, any muscle in the body can be affected, including the tongue. Different muscles can be involved at the same time or successively. The vigor of the affected muscles generally increases. Myotonic phenomenon appears in 50% of cases. Normal reflexes.

Hyperhidrosis of the limbs or affected site may frequently be observed. There is no visceral or skeletal shot.

Paraclinical examinations

Electromyogram. Normal.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Adams, R.D.: D. Denny-Brown and C. M. Pearson: Diseases of the muscle. New York, Harper Row, 1962.

Friedreich, N.: "Ueber congenitale halbseitige Kophypertrophie." Virchow's Arc. Path. Anat. 28:474-481. 1863.

MEGACONIAL MYOPATHY SYNDROME

Synonymy

S. of mitochondrial or lipid storage myopathy.

Syndromography

Clínica

Slowly evolving congenital myopathy that appears in childhood, but has been observed at older ages, including towards mid-adult life. It is characterized by hypotonia and weakness of the proximal muscles with difficulty walking and ascending stairs. Diminished or absent tendon reflexes, no myotonia or fasciculation, no loss of sensation or coordination.

Paraclinical examinations

Muscle biopsy. Presence of giant mitochondria (100 times larger than normal) with abnormal "torn fibers with red" so called because of their appearance with the staining of the muscle sample.

Syndromogenesis and etiology

Alterations of the essential enzymes for lipid metabolism contained in the mitochondria that give rise to an abnormal accumulation of lipid bodies in the muscle fibers.

The study of mitochondrial DNA (mtDNA) of affected people has shown in muscle mtDNA populations that there are some normal and others with deletion of a 7 kilobase mtDNA segment. This observation demonstrates that the etiology of this syndrome is due to a defect in the mitochondrial genome, with maternal transmission, with the severity of the syndrome corresponding to the mtDNA defect and its metabolic correlation and to the random distribution of normal and abnormal mitochondria in maternal gametogenesis.

Literature

Holt, I.J. et al.: "Deletions of muscle mitochondrial DNA in patients with mitochondrial myopathies." Nature, 331:717-719, 1988.

Shy, GM.; N.K. Gonatas and M. Pérez: Two childhood myopathies with abnormal mitochondria: I megaconial myopathy, II pleoconial myopathy". Brain, 89:133-158, 1966.

NEMALINE MYOPATHY SYNDROME

Synonymy

Rod's myopathic S.

Syndromography

Two types are described: early installation and late installation.

1. EARLY INSTALLATION

Clinic

It is installed at birth. There is delayed motor development, proximal limb weakness, reduced muscle volume, muscle hypotonia, areflexia or normal reflexes in some cases. Associated malformations: high-arched palate, pes cavus, kyphosis or scoliosis.

Paraclinical examinations

Muscle biopsy. 20-40% of the fibers are affected, which contain a variable amount of filamentous formations. The nuclei of the injured cells are vesicular and have a prominent nucleolus.

Blood. Creatinine index. Increased

Glutamic oxalacetic transaminase. Normal.

Amino acids. normal excretion

Electromyogram. Decrease in the duration of the Tencials.

Syndromogenesis and etiology

Of genetic etiology, the existence of autosomal recessive and dominant inheritance has been suggested.

Type 2 fiber deficiency could explain the basis of patients' inability to perform gross motor activity.

2. LATE INSTALLATION

Clinic

Both sexes are affected. It is established in the fourth to sixth decades in previously healthy subjects without developmental abnormalities. The installation is gradual in the distal parts of the legs or pelvis, progressing towards the proximal and then distal parts of all the limbs, also affecting the neck flexors.

Physical examination. There is muscle hypotonia. Reflexes generally decreased or normal.

Syndromogenesis and etiology

Unlike the previous type, no cases have been reported in families.

The etiology is unknown.

Literature

Shy, G.M.; W.K. Engel, et al.: "Nemaline myopathy, a new congenital myopathy." Brain, 86:793-810, 1963.
Wallgren-Petterson, C., et al.: "Genetics of congenital nemaline myopathy: a study of 10 families." J. Med. Genet.. 27:480-487, 1990.

MORTON SYNDROME (T)

Synonymy

S. of the short first metatarsal. S. of Morton's triad.
S. of metatarsalgia. S. of digital neuroma. S. of intermetatarsal neuroma.

Syndromography

Clínic

Predominates in women. It is unilateral or often bilateral.

It is characterized by recurrent burning-type pain between the third and fourth metatarsal spaces that radiates to adjacent parts of the foot. The discomfort can persist even at rest and prevent sleep. There is excessive foot fatigue.

There is also pain in the longitudinal arch with radiation to the calf. The pain begins after walking or after certain standing positions. It is relieved with rest.

Physical examination. The foot is abnormal in appearance. Careful examination reveals that the metatarsus may be slightly displaced upward and toward the midline and has abnormal mobility; It is also covered by soft, smooth skin, while the heads of the second and third metatarsals are prominent and a hard callus can be located under them.

The pressure exerted between the third and fourth spaces of the metatarsal heads reproduces the pain.

Paraclinical examinations

Radiology. It reveals shortening of the first metatarsal and secondary changes.

Syndromogenesis and etiology

There are congenital malformations of the first metatarsal.
Acute symptoms are secondary to synovitis of the second and third metatarsal and cuneiform joints.
Chronic symptoms are due to hypertrophic osteoarthritis.

The condition is caused by compression of the digital nerve by the metatarsal head. It can also be caused by neurofibromas or angioneurofibromas of the median plantar nerve.

Literature

Bingham, R.: "Painful feet; congenital insufficiency of first metatarsal segment as cause among soldiers recently inducted into the army." J.A.M.A., 124:283-286, 1944.
Morton,T.: "Metatarsus ataviscus. The identification of a distinct type of foot disorder". J. Bone Joint Surg., 9:531-544, 1927.
Morton, T.: Foot disorders in general practice." J.A.M.A., 109:1112-1119, 1937.

RECTUS MUSCLE SYNDROME

Synonymy

S. of hematoma of the rectus muscle.

Syndromography

Clinic

It occurs in middle-aged patients, more common in women. Sudden, severe pain appears on both sides of the abdominal midline, always below the level of the navel, moderate increase in temperature, prostration, vomiting, shock. In other cases, a mass without acute symptoms slowly develops in the paramedian abdominal region.

Physical examination. A painful mass is noted in the affected site, which always remains fixed, sometimes ecchymotic, and does not disappear with the voluntary contraction maneuver of the abdomen, tachycardia. If the patient sits the mass is less painful. There may be localized tonic contractions and absence of generalized abdominal rigidity or contracture.

Paraclinical examinations

Tumor puncture. Aspiration.

Laboratory tests. Negatives

Laparoscopy. It allows us to demonstrate that it is a tumor of the wall with integrity of the abdominal cavity.

Surgical exploration. It is not justified, the treatment must be conservative.

Syndromogenesis and etiology

It is due to a sudden muscle contraction during an effort such as coughing, sneezing, vomiting, sudden movements with the abdominal muscles.

It is frequently associated with infections, pregnancy, poisoning, which causes weakness of the abdominal wall, degeneration of the vessels of the wall and/or blood dyscrasia.

nea.

Literature

Hippocrates (quoted by Payne). Payne, R.L.: "Spontaneous rupture of the superior and inferior epigastric arteries within the rectus abdominal sheath." Ann. Surg., 108:757-768, 1938.
Merendino, K.A. and T.W. Jones: "The deep epigastric artery: rectus muscle syndrome". Amer. J. Surg., 103:159-169, 1962.

NAFFZIGER SYNDROME

Synonymy

Adson S. of Coote. from Haven.

Syndromography

Clinic

In this syndrome, exercise triggers the symptoms and rest reduces them. There is pain from the neck to the hand or above the deltoid with extension to the arm, mostly to the elbow. It is accompanied by paresthesia and weakness of the affected arm, occasionally hyperesthesia in painful areas. Vasomotor alterations are frequently present.

Physical examination. The Adson maneuver induces symptoms. There is decreased or absent reflexes in the affected arm and a painful point in the scalene muscle.

Paraclinical examinations

Radiology. It should be performed in the neck and shoulder regions in different projections.

Syndromogenesis and etiology

Naffziger syndrome was considered in the past as a consequence of spastic irritability and scalene hypertrophy that compresses the neurovascular branch. Today this mechanism is rejected and it is proposed that the improvement of symptoms after a tenotomy is produced by the release of a passive mechanism and postoperative rest, rather than the correlation with a pathogenetic mechanism.

Literature

Jackson, R.: The syndrome of the cervical nerve root compression.
In Hollander, J.L.: Arthritis and allied conditions. ed 7, Philadelphia, Lea & Febiger, 1966.
Naffziger, H.C. and W.T. Grant: "Neuritis of the brachial plexus mechanical in origin. The Scalenus syndrome." Surg. Gynec. Obstet.. 67:722-730, 1938.

PATELLAR PLEXUS NERVE NEURITIS SYNDROME

Synonymy

S. of traumatic prepatellar neuralgia.

Syndromography

Clínic

Symptoms of electric shock occur when well-defined unilateral or bilateral trigger zones are lightly stimulated. The sensation appears, disappears and recurs without reason.

rent.

Syndromogenesis and etiology

It can be caused by a small and forgotten trauma.

An anatomical accident outside the infrapatellar branch of the saphenous or other nerve of the prepatellar plexus, vulnerable to mechanical trauma or ischemia produced by movement.

The etiology is unknown.

Literature

Wartenberg. A.: *Digitalgia paresthetica and gonyalgia paresthetica". Neurology, 4:106-115, 1954.

NIELSEN SYNDROME I

Synonymy

S. of neuromuscular exhaustion. S. of generalized neuromuscular exhaustion.

Syndromography

Clinic

This syndrome develops subacutely after great effort during a period of euphoria. The degree of overwork is variable and depends on age and other factors. There is a feeling of deep exhaustion throughout the body, more severe for the muscles subjected to effort. There is pain, tremor and atrophy of the affected muscles.

Paraclinical examinations

Cerebrospinal fluid. Normal.

Electromyography and studies of thyroid function.

They must be performed to rule out known diseases.

Syndromogenesis and etiology

There are metabolic disorders due to great effort.

The etiology is unknown.

Literature

Nielsen, J.M.: Subacute generalized neromuscular exhaustion syndrome." Bull. Los Angeles Neurol. Soc., 5:128-130, 1940.
Nielsen, J.M.: "Subacute generalized neuromuscular exhaustion syndrome: report of 3 cases." Qual. Med., 66:338-340, 1947.

OLLIER SYNDROME

Synonymy

S. of enchondrosis. S. of multiple enchondrosis.

Syndromography

Clinic

Patients present various disorders of the hands, feet, femur, fibula and pelvis, generally unilateral. Occasionally they suffer from pathological fractures.

Physical examination. There is deformity of the bones mentioned-

two.

Paraclinical examinations

Skeletal radiology. Shows radiotransparent zones

relatives.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Ollier, M.: "Sur une nouvelle affection: la dyschondroplasie."
Rev. of Chir. Paris, 21:396-398. 1900.

OPPENHEIM SYNDROME

Synonymy

S. of congenital amyotonia. S. of congenital myoatonia.

Syndromography

This syndrome has been subdivided into different syndromes, since it represents a heterogeneous group:

1. Lange type muscular dystrophy.
2. Batten-Turner muscular dystrophy.
3. Benign congenital hypotonia syndrome.

We will describe the latter as a group of non-progressive congenital hypotonias of muscular origin, generally benign. Its forms are: central core, nemaline myopathy, mega and/or pleoconial myopathy, myotubular.

Clínica

In the prenatal phase the mother feels few fetal movements, in the neonatal phase there is generalized hypotonia, strange postures, legs in an "open book" when the newborn is face up.

Facies: hanging jaw. There is difficulty in feeding
tation.

Flat chest, with breathing difficulties if the diaphragm is involved in the myopathy. Tendon reflexes are slow and weak. Intelligence is normal, but there is delay
engine.

Paraclinical examinations

Electromyogram. To exclude neural atrophies.

Muscle biopsy. It allows you to recognize the different shapes. Many muscle fibers present degeneration in their center with a lack of phosphorylase. Large mitochondria. The nuclei are grouped concentrically. Damaged muscle fibers lose strength.

Blood. The determination of the enzymes aldolase, transaminase and creatine phosphokinase is normal.

Syndromogenesis and etiology

The term congenital amyotonia is reserved for conditions in which the primary abnormality resides in the muscles and is essentially nonprogressive.

The central core and nemaline forms are sporadic or transmitted by autosomal dominant inheritance.

Literature

Becker, P.E.: Myopathien in human genetics. t 3, p. 411, 1964.

Byers, P.E. and B.C.H. Banker: "Infantile muscular atrophy." Arch. Neurol., 5:140, 1961.

Oppenheim, H.: Üeber allgemeine und localisierte Atonie der muskulatur (myatonia) im frűehen Kindersalter". Mschr. Psychiat. Neurol., 8:232-233, 1900.

Zellweger, H.: The flappy infant. London, W. Heineman, 1969.

AURICULAR OSTEODYSPLASIA SYNDROME

Syndromography

Clinic

It affects both sexes. There is slight limitation of elbow movements, produced by a dysplasia that ranges from moderate dysplasia to both anterior and posterior dislocation (often bilaterally asymmetric). Hip dysplasia Occasionally bilateral that occurs only in women in the affected family. Masculine appearance of the torso, broad shoulders, wide base of the neck, horizontal clavicles, prominence of the scapula, shortening of the metacarpals in some patients. Normal nails, teeth and hair. There is ear dysplasia: elongation of the lobe accompanied by a small posterior lobe.

Height below 50 percentiles in height.

Paraclinical examinations

Bone radiology. Check the alterations reported in the clinic.

Syndromogenesis and etiology

This syndrome is transmitted by autosomal dominant inheritance.
nante.

The etiology is genetic.

Literature

Beals, R.K.: "Auriculo-osteodysplasia, a syndrome of multiple osseous-dyplasia, ear abnormaly, and short stature." J. Bone Joint Surg., 49A: 1541-1550, 1967.

MIGRATORY OSTEOLYSIS SYNDROME OF THE LOWER LIMBS

Syndromography

Clinic

It appears after moderate trauma or spontaneously. It is characterized by migratory soft tissue pain.

Physical examination. There is edema of the legs, erythema, dry skin, dilated superficial veins.

Segmental episodes may appear in other areas of the opposite leg without an initial cause.

Paraclinical examinations

Radiology. Regional and migratory osteolysis.

Syndromogenesis and etiology

The etiology is unknown.

Bibliography

Duncan, H.; B.Frame; H.M. Frost, et al.: "Migratory osteolysis of lower extremities", Ann. Intern. Med., 66:1 165-1173, 1967.

Hunder, GG. and P.J. Kelly: "Roentgenologic transiet osteoporosis of the hip, a clinical syndrome?" Ann. Intern. Med., 68:539-552, 1968.

OSTEOMYELITIS SYNDROME OF THE SPINE AND URINARY TRACTUS INFECTION

Syndromography

Clinic

It appears in men in the fifth and seventh decades. It follows a urinary tract infection. There is low back pain and meningeal signs.

Paraclinical examinations

Blood culture, urine culture and culture of osteolytic lesions. They demonstrate an infection to different germs.

Spine radiology. Locate the lesion and show its extent.

Syndromogenesis and etiology

The most frequently found germs are:
Escherichia coli, Proteus, Pseudomona.

bibliography

Carson, H.W.: “Acute osteomyelitis of the spine.” Brit. J. Surg.. 18:400-408, 1931.

Sierra, M.A.: FJ. Luparello and JR Lewin: Vertebral osteomyelitis and urinary tract infection." Arch. Intern. Med., 108:128-131, 1961.

PAGET SYNDROME

Synonymy

S. of osteitis deformans. St. of Pozzi

Syndromography

Clinic

This syndrome is more common in men but more severe in women. The installation is insidious. It begins after age 40 and is frequently asymptomatic. Headache episodes may occur. Constant pain in one knee. Deafness.

Physical examination. Widening of the cranial vault is noted. Bone deformities, shortened height, kyphosis.

Paraclinical examinations

Radiology. Bone resorption, fibrotic changes, disorganization of trabeculae, bone deformities.

Blood. Hyperphosphatemia, increased alkaline phosphatase, hypercalcemia.

Urine. Hypercalciuria.

Syndromogenesis and etiology

It has been suggested that Paget's disease is a slow virus infection.

The etiology is unknown.

Literature

Harvey, L. et al.: "Ultrastuctural features of the osteoclast from Paget's disease of bone in relation to a viral aetiology." J. Clin. Pth., 35:771-779, 1982.

Howell, D.: Metabolic bone diseases. In Hollander, J.L.: Arthritis and allied conditions, ed. 7, Philadelphia, Lea & Febiger, 1966.

Paget, J.: "On a form of chronic inflammation of bones (osteitis deformans)". Med. Chir. Trans (London), 60:37-63, 1877.

ANTERIOR CHEST WALL SYNDROME

Synonymy

S. of Prinzmetal. S. of Massumi.

Syndromography

Clínica

There is pain in the anterior region of the chest associated with discomfort when pressure is applied with the fingers, which is not relieved by rest, ingestion of food, or nitroglycerin.

Five types are described according to their location:

1. Pectoralis major syndrome (more common on the left side). The pain is located in the upper half of the chest in the second and third costal regions. Sometimes the pain can radiate to the arm. It may exist in association with heart disease.
2. Inframammary syndrome (more common in women, located on the left side). The pain occurs under the breast, in the nedioclavicular line of the sixth or seventh intercostal space. It may coexist with gastrointestinal pathology. Responds to antispasmodics and antacids.
3. Marginal costal syndrome. It occurs on both sides of the chest. The pain is located at the margin of the eighth, ninth and tenth ribs and at their attachment site. Responds to specific treatment.
4. Costochondral junction syndrome. There is pain in the upper chest at the costochondral junction.
5. Xiphoid process syndrome. It has already been described, the pain is located exactly on the xiphoid.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Long, C.: "Myofascial pain syndromes." Henry Ford Hosp. Med. Bull., 4:102-106, 1956.

Prinzmetal, M. and R. Massumi: "The anterior chest wall syndrome- chest pain resembling pain of cardiac origin." J.A.M.A., 159:177-184, 1955.

TENNIS LEG SYNDROME

Synonymy

S. of the rupture of the calf muscles.

Syndromography

Clinic

It sets in suddenly with an audible pop in the leg during strong exercise, accompanied by intense pain in the calf that can extend to the popliteal space and is increased by dorsiflexion of the foot.

Syndromogenesis and etiology

This syndrome is due to a sudden muscle contraction with rupture of one of the calf muscles during violent exercise.

Literature

Hollander, J.L.: Arthritis and allied conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.

LONG LEG SYNDROME

Syndromography

Clinic

Around 15% of the population has a leg length difference of 1 cm or more compared to the other. Patients are generally asymptomatic or may have the same symptoms as those with a large difference.

Patients report pain in the buttock, hips and sometimes in the lower back, knees or occasionally in the calves.

Physical examination. A difference is noted in the length of the legs with an increase in one of them.

Paraclinical examinations

Radiological study. It allows you to check the alterations produced by the difference in length, at the level of the hips and spine.

Syndromogenesis and etiology

The etiology is unknown

Literature

Gofton, J.P. and GE. Trueman: "Unilateral idiopathic osteoarthritis of the hip." Dog. Med. Ass., 97:1 129-1132, 1967.

PIRIFORMES SYNDROME

Syndromography

Clínic

This syndrome occurs in patients with a history of trauma to the sacroiliac or gluteal region (often forgotten).

The patient reports pain in the sacroiliac joint, in the large sciatic notch and in the piriformis muscle, radiating to the leg, which causes difficulty walking.

There is an acute exacerbation of chronic pain after lifting weights.

Physical examination. An elongated mass is palpated over the piriformis muscle during the pain attack. Positive Laségue sign. Gluteal atrophy according to the chronicity of the process.

Paraclinical examinations

Radiological studies. They allow us to exclude other processes, lumbosacral and the hip joint.

Syndromogenesis and etiology

There is an injury to the piriformis muscle that affects the sciatic nerve, in those cases where there is a relationship between this muscle and the sciatic nerve.

Literature

Robinson, D.R.: "Pyriformis syndrome in relation to sciatic pain." Amer. J. Surg., 73:355-358, 1947.

Yoemans, W.: "The relationship of arthritis of the sacroiliac joint to sciatica." Lancet, 2:1119-1 122, 1928.

RELAXING POLYCHONDRITIS SYNDROME

Synonymy

S. of chondromalacia. S. of chronic atrophic perichondritis.

S. of diffuse perichondritis. S. of polychondropathy. S. von Meyenburg.

Syndromography

Clínic

80-90% of patients suffer attacks in the ears and nose, and report spontaneous pain or palpation and swelling of the cartilaginous portion of the ear. Repeated episodes can cause the ears to be lax and pendulous.

Attacks are accompanied by recurrent episodes of malaise and low fever, sometimes painful polyarthritis. It is characterized by episodes of inflammation followed by destruction of the nasal cartilage, which causes deformity of the nose.

Dyspnea and respiratory distress frequently occur due to the narrowing of the trachea and bronchial tubes secondary to respiratory failure and lung infections.

recurring.

There may be interference with hearing, obstruction of the Eustachian tube with secondary otitis media, and deafness.

There is vertigo, ataxia, nausea and vomiting.

Conjunctivitis, episcleritis, iritis are common.

Ulcerations in the mouth and genitals. Episodes of non-deforming polyarthritis in large and small joints.

Aneurysms and thromboses may occur, as well as focal proliferative glomerulonephritis.

Physical examination. Saddle nose. Wheezing in both lung fields. 30% of patients present scleritis, conjunctivitis, iritis.

All cartilaginous joints may be affected (pubic symphysis, sternal manubrium, vertebral).

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation during attacks. Normocytic normochromic anemia, positive rheumatoid factor and antinuclear antibodies in some cases, sometimes circulating immune complexes.

Rheumatoid arthritis tests. Negatives.

Urine. Increased excretion of mucopolysaccharides.

Cartilage biopsy. Inflammatory changes are observed followed by cartilage destruction.

Radiology. Bone Survey. Calcifications in cartilage of the nose, larynx and trachea.

Bronchography. You can check tracheal stenosis.

Syndromogenesis and etiology

It is an inflammatory disorder of unknown origin that affects cartilaginous structures, cardiovascular system, eyes and ears.

There is loss of cartilage glycosaminoglycans.

At the edge of destruction, lymphocytes and plasmacytes are observed. Invasion of cartilage by granulation tissue.

Cellular and humoral immunity mechanisms intervene in tissue injury.

Patients present cartilage destruction. Respiratory distress is produced by the narrowing of the trachea and bronchi.

The etiology is unknown.

Literature

Hernan, JH.: "Polychondritis, in textbook of Rheumatology" W.N. Kelley et al., Philadelphia, Saunders, 1985. pp. 1458-1467.

Hewe, R.: "Relapsing polychondritis." Brit. Med. J., 4:661-662, 1967.

Jaksch-Wartenhorst, R.: “Polychondropathia”. Vienna. Arch. Inn. Med., 6:93-100, 1923.

POLYMYALGIA RHEUMATICA SYNDROME

Synonymy

S. of rheumatoid anarthritis. S. de Forestier-Certonciny.

Syndromography

Clinic

It affects younger people. The installation can be acute or insidious. There is pain and stiffness of the shoulders and hips, which is occasionally severe and disabling. There is anorexia, weight loss and fever. It is frequently associated with Horton syndrome.

Physical examination. Muscle rigidity is noted. There is absence of joint deformity.

Paraclinical examinations

Blood. Accelerated erythrocyte sedimentation. L.E. cells and negative rheumatoid factor.

Biopsy of skin, mucosa and muscle. Negatives.

Syndromogenesis and etiology

In general, it is part of the collagen and granulomatous syndromes, without being able to be easily separated from other similar ones; especially it is considered very associated or coexisting with giant cell arteritis previously known as temporal arteritis or Horton syndrome and responds to the same treatment.

The etiology is unknown.

Literature

Barber, H.S.: "Myalgic syndrome with constitutional effects; polymyalgia rheumatica". Ann. Rheumat. Dis., 16:230-237, 1957.

Bell, W.R. and HF. Klinefelter: "Polymyalgia rheumatica." Johns Hopkins Med. J., 121:175-187, 1967.

Bruce, W.: College of GP. Research Newsletter, 9:157, Brit. Med. J., 2:811, 1888.

IDIOPATHIC POLYMYOSITIS SYNDROMES

Synonymy

S. of polymyositis.

Syndromography

Clinic

The common symptoms for all groups are: symmetrical muscle weakness (more frequently affecting the proximal muscles). In group 1, the shoulder muscles are affected with myalgias that cause an increase in muscle consistency. It is frequently associated with Raynaud's phenomenon and dysphagia.

Physical examination. Loss of the Achilles reflex in severely affected patients.

Four groups are described:

Group 1

Acute, more common in young people (1-20 years).

Subacute. It affects childhood.

ChronicI. Early adulthood.

Chronicle II. Older adults. Late stage (muscular dystrophy).

Group 2

Polymyositis is dominant. Associated connective tissue disease plays a minor role.

Group3

Poliomyositis is minor. The associated connective tissue involvement has a major role.

Group 4

Polymyositis is associated with carcinoma.

Paraclinical examinations

Muscle biopsy. It presents the typical lesions of polymyositis: acute or subacute degeneration of muscle fibers with variable inflammatory alterations.

Electromyogram. When it is typical, it shows the following three elements: low-amplitude polyphasic motor unit potentials, pseudomyotonic high-frequency tracing, and spontaneous fibrillation with positive sharp waves (sawtooth type) in the muscle at rest.

Blood. L.E. cells, rheumatoid factor. Erythrosis- normal or elevated sedimentation. Increase, not constant, of the aldolase, TGO, LDH and CPK (creatine phosphokinase). Elevated serum myoglobin in 75% of patients.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Adams, R.D.; D. Denny-Brown and C.M. Pearson: Diseases of the Muscle. Harper & Row, New York, 1962.

Stevens, M.B.: Polymyositis (Dermatotomyositis), In A.S. Cohen (Ed.). Rheumatology and Immunology. Grune & Stratton, New York, 1979.

Wagner, E.: "Fall einer seltnen Muskeltrankheit." Arch. Heilkunde, 4:282-283, 1863.

POSTURAL SYNDROME

Syndromography

Clínica

There are two types: functional and structural.

The functional type prevails in children and is manifested only by fatigue and slight back pain. The structural type occurs at all ages and back pain is more intense.

Physical examination. There is an increase in cervical lordosis, decreased motility in the dorsal column. The lumbar lordosis curve is exaggerated. The abdominal muscles are flaccid, the pelvis tilted forward and the hip flexors are shortened. There is an increase in the lumbosacral angle.

Paraclinical examinations

Radiology. Spinous processes and facets in contact with each other. Secondary degenerative changes of the bones.

Syndromogenesis and etiology

Pregnancy and obesity are frequently determining or precipitating factors.

Literature

Kerr, W.J. and J.B. Lagen: "The postural syndrome related to obesity leading to postural emphysema and cardiorespiratory failure." Ann. Intern. Med., 10: 569-595, 1936.

Kuhns, J.G: Posture and its relationship to orthopedic disabilities. Ann. Arbor, Edward Bros, 1942.

PSEUDOACHONDROPLASIA SYNDROME

Syndromography

Clínica

This syndrome affects both sexes. Patients present short stature during adolescence. On physical examination, other manifestations may be found, such as:
osteoarthritis of the hips and also occasionally, knees and shoulders.

Paraclinical examinations

Radiology. In children: there is fragmentation of the epiphyseal ossification centers. In adults: changes in the spine. Osteoarthrosis.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

There is fragmentation of epiphyseal ossification centers.

The etiology is unknown.

PSEUDOGOUT SYNDROME

Synonymy

Calcium pyrophosphate and hydroxyapatite deposition disease. S. of chondrocalcinosis. S. of chondrocalsinovitis.

Syndromography

Clínic

In 25%, acute attacks (pseudogout) may appear that set in quickly and reach their maximum intensity in 12-36 hours.

More common in women over 40 years of age. Symptoms range from mild arthralgia to acute arthritis similar to gout attacks (which last from one day to eight weeks). The most frequently affected joints are:

knees, hands and feet; 20% have polyarthralgias.

It is characterized by: erythematous joints, with increased volume and very painful. It is usually limited to a single joint. The most common site is the knee, although ankles, wrists, elbows, hips, and the cervical and lumbar spine may be affected. It can attack the metatarsophalangeal joint of the big toe, as in gout. These symptoms may subside in one or two weeks.

Pseudorheumatoid disease appears in a smaller percentage of patients, characterized by attacks in multiple joints, lasting weeks or months.

There is morning stiffness and fatigue.

Another group (50%) adopts the chronic form, with multiple joints taken: knees, wrists, hips, shoulders, elbows and ankles, symmetrical in their location.

Destructive lesions may arise in one or more joints and simulate neuropathic arthropathy.

Physical examination. Variable degree of inflammation of the affected joints.

Paraclinical examinations

Study of joint fluid. There is a large number of polymorphonuclear cells. Presence of calcium pyrophosphate crystals, which can be visualized using polarized light.

Radiology. There are calcifications in the hyaline cartilage of the joint, with the appearance of dense fine lines on the knees, elbows, and wrists. Calcifications in tendons, frequently the Achilles and supraspinatus tendons.

Synovial fluid aspiration. Calcium crystals appear intracellularly and extracellularly.

Arthroscopy and Biopsy.

Blood. Hyperuricemia, hyperglycemia or impaired glucose tolerance test. Erythrocyte sedimentation usually accelerated. Serum calcium and phosphorus almost always normal.

Normal alkaline phosphatase.

Syndromogenesis and etiology

The deposition of calcium pyrophosphate dihydrate crystals in the joints is known as joint disease.

pyrophosphate deposition and is characterized by acute and chronic inflammatory arthropathy. The acute form is called pseudogout.

Metabolic defect with precipitation of calcium pyrophosphate crystals in synovial fluid, cartilage and periarticular structures.

Literature

McCarthy, D.J.; N.N. Kohn and J.S. Faires: "Significance of calcium phosphate crystals in synovial fluid of arthritic patients: the 'pseudodogout syndrome'. Clinical aspects". Ann. Int. Med., 56:711-737. 1962.

McCarthy, D.J.: "Pseudogout and pyrophosphate metabolism." In Advances in Internal Medicine. G. H. Stolleman, Year book.
1980.

Moskowitz, R.W. and D. Katz: *Chondrocalcinosis and chondrocalsynovitis (pseudogout syndrome)." Amer. J. Med., 43:322-334, 1967.

BAKER'S QUEST SYNDROME

Synonymy

S. of popliteal cysts.

Syndromography

Clinic

This syndrome occurs at any age. The patient reports moderate pain and stiffness in the knee, usually unilateral.

Physical examination. There is edema of the popliteal space that may extend to the Achilles tendon.

Paraclinical examinations

Transillumination. Reveals the presence of cysts.

Syndromogenesis and etiology

It can be produced by any cause that increases the tension of the synovial fluid. There is posterior herniation of the knee capsule, enlargement of the semimembranous bursa, and leakage of synovial fluid from the knee into one of the bursae.

The etiology is unknown.

Literature

Baker, W.M.: "Baker's cyst; formation of abnormal synovial cysts in connection with joints." Med. Classics, 5:805-820, 1941.

Hollander, J.L.: Arthritis and allied conditions. Philadelphia, Lea & Febiger, 1966.

EXERCISE RHABDOMYOLYSIS SYNDROME

Syndromography

Clínic

This syndrome can occur at all ages, but it occurs more frequently in young men, enrolled in military service or in athletic circles. The episodes occur after performing stressful calisthenics without prior preparation. Patients report severe myalgia of stressed muscles that lasts from a few days to a month, with recovery in one or several months. The patient reports passing dark brown urine between 24 and 38 hours after exercise. It is almost always preceded by discomfort and oliguria.

Paraclinical examinations

Urine. Myoglobinuria and proteinuria.

Blood. TGO, CPK, aldolase and lactic dehydrogenase are increased.

Syndromogenesis and etiology

It is due to rhabdomyolysis after heavy exercise.

Literature

De Lange, C.D.: "Myoglobulin and myoglobinuria." Acta Med. Scand., 124:213-226, 1946.

Smith, R.F.: "Exertional rhabdomyolysis in naval officer candidates." Intern Arch. Med. 121:313-319, 1968.

RECKLINGHAUSEN SYNDROME I

Synonymy

S. of generalized osteitis fibrosa. S. of primary hyperparathyroidism. S. of osteitis fibrosa cystica. S. of Engel-Recklinghausen.

Syndromography

Clínica

Manifestations of nephritis Symptoms of primary or secondary hyperparathyroidism. Arthralgias. Appearance of fractures. Curved long bones and deformities of the spine and thorax occur. Progressive decrease in motility accompanied by muscle hypotonia. Spontaneous fractures may occur. Nephrolithiasis and nephrocalcinosis are common in its evolution.

Paraclinical examinations

Bone radiology. Formation of cysts in areas with bone rarefaction. The bone marrow is replaced by fibrous tissue.

Ultrasound. There is kidney stones.

Blood. Serum calcium and alkaline phosphatase are increased. Phosphorus is decreased.

Urine. Calcium is decreased. Phosphorus, increased.

Study of kidney and parathyroid function.

Syndromogenesis and etiology

This syndrome occurs in chronic nephritis, and in primary hyperparathyroidism and with increased osteoblastic and osteoclastic activity.

Literature

Engel, G.: "Ueber einen Fall cystoider Entartung des gesamten Skeletts. Giessen, 1864.

Trueta, J.: Studies of the development and decay of the human frame. Philadelphia, Saunders, 1968.

Von Recklinghausen, F.: "Untersuchungen über Rachitis und Osteomalacie." Jena, G. Fisher, 1910.

WEAK KNEE SYNDROME

Syndromography

Clinic

Discomfort in the knee when the patient is standing. The knee joint is relaxed and there is atrophy of the quadriceps femoris.

Paraclinical examinations

Radiology. Diffuse demineralization of the bones that make up the knee.

Syndromogenesis and etiology

It is caused by previous damage to the knee or soft muscles to which the aforementioned atrophy of the quadriceps is associated.

SCHEUTHAUER-MARIE-SAINTON SYNDROME

Synonymy

S. de Scheuthauer. S. of Hulkerant anosteoplasia. S. of cleidocranial dysostosis. S. of mutational dysostosis. S. of osteodental dysplasia. S. de Marie-Sainton.

Syndromography

Clínic

This syndrome is characterized by its family history, as well as by independent cases. Patients present varying degrees of aplasia of the clavicles that lead to rare mobility of the shoulders. It is accompanied by neurological and vascular symptoms produced by the compression of the rest of the clavicle that exists.

Already in the newborn, respiratory distress is seen as a result of the thoracic deformity. Large and wide fontanels, sutures with delayed closure. Saddle nose, delayed teething, supernumerary teeth, narrowing of the chest, abnormal gait. Small facial bones, absent or small mastoid air cells.

Hypertelorism, kyphosis, scoliosis, lordosis, and spina bifida may occur.

Pathological fractures are common. Epilepsy, schizophrenia and mental retardation are frequently present.

Paraclinical examinations

Radiology. A bone survey allows us to appreciate the indicated alterations.

The following alterations are summarized: brachycephalic skull with an increase in biparietal diameter, wide sutures, the anterior fontanel open until adulthood, posterior occipital synchondrosis that persists until 4-5 years of age, hypoplasia of the sphenoid, prognathism, short ribs, aplasia of the clavicle, small scapulae, coxa vara or coxa valgus, deformity of the femoral head, bone alterations iliac and other alterations.

Syndromogenesis and etiology

The etiology is genetic and is transmitted by autosomal dominant inheritance with marked variations in genetic expression. A type has been described that is accompanied by micrognathia, which presents autosomal recessive inheritance.

Literature

Chitayat, D.; K.A Hodgkinson: “Intra familial variability in cleidocranial dysplasia: a three generation family.” Am. J. Med. Genet., 42:298-303, 1992.

Fauré, C. et al.: Cleidocranial dysplasia, in Kaufman, H.J. Progress in Pediatric Radiology. vol 4, Basel-Karger, 1973, p. 201.

Foriano, M.: "Cleidocranial dysostosis." Am. J. Med. 33:792, 1962.

Koischwitz. D. et al.: "Dysplasia cleidocraniatis." Forts chr. Rontgenstr., 119:438, 1973.

Marie, P.; P. Sainton: "Observation D'ydrocephalie Hereditaire (Pére et Fils) Parvice de Development du crane et ou cerveau." Bull. Mem. Soc. Méd. Hop. Paris 14:706. 1897.

Marie, P.; P. Sainton: "sur la dysostose cléido-cranienne hereditaire." Bull. Mem. Medical Soc. Hop. Par., 15:436, 1898.
Scheuthauer, G.: 'Kombiantion rudimentarer. Schlussel beine mit Anomalien des Schadels beim erwachsen Menschen". All. Weir. Med. Ztg., 16, 293, 1929.

SCHMID SYNDROME

Syndromography

Clinic

Established early in childhood, it affects both sexes.

Physical examination. Delayed growth, bowed legs, short shafts, inability to fully extend the fingers, normal or short trunk.

Paraclinical examinations

Radiology. More marked metaphyseal dysplasia in the lower limbs, particularly in the ankles and knees. Coxa Stick.

Syndromogenesis and etiology

Scars appear in the metaphyses of bones. Shortening of bones.

The etiology is unknown.

Literature

Miller, S.M. et al.: "Roentgen observations in familial metaphyseal dysostosis." Radiology, 83. 665, 1964.
Rosenbloom, A. et al.: "Tite natural history of metaphyseal dysostosis." J. Pediatr., 66:857, 1965.
Schmid, E.: "Beitrag zur Dysostosis enchondratis metaphysaria." Monatsschr. Kinderth.. 97:393-397, 1949.
Tay Bi, H.; AD Mitchell and G. D. Friedman: "Metaphyseal dysostosis and the associated syndrome of pancreatic insufficiency and blood disorders." Radiology: 93:563, 1969.

SECRETAN SYNDROME

Syndromography

Clinic

After a trauma, physical examination reveals restriction of flexion of the fingers of the hand except the thick finger, or edema of the dorsal region of the metacarpals and base of the fingers.

If there is trauma to the foot, there is post-traumatic edema of the dorsum of the toes.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Grobmyer, A.J.; J.M. Bruner and L.R. Dragstedt: "Closed lymph phanoplasty in Secretan's disease." Arch. Surg., 97:81- -83.
Secretan, H.: "Oedeme dur et hyperplasie traumatique du metacarpe doral." Rev. Med. Suisse Rom., 21:409-416, 1901.

SHY-MAGGE SYNDROME

Synonymy

S. of the central core. S. of central core.

Syndromography

Clinic

It affects both sexes. It is installed during the first year of life. There is proximal muscle weakness. Delay in the development of gait. Some patients take 5 years to start walking. Normal mental development.

Physical examination. Symmetrical hypotonia of the proximal muscles with predominance in the lower limbs, in relation to the upper limbs. There are no fasciculations or myotonia.
Tendon reflexes are normal or decreased.

Paraclinical examinations

Muscle biopsy. Alteration of striated myofibrils with disorganization of the normal striation. Atrophy of type I muscle fibers, central core and central area pale, with variable color granules.

Biochemical studies of muscle metabolism.
Creatinine index. Increased.

Glutamic oxalacetic transaminase. Normal.

Electromyography. Decreasing duration of potential action.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance and is an example of genetic heterogeneity.
The etiology is genetic.

Literature

Shy, G.M. and K.R. Magee: "A new congenital non-progressing myopathy." Brain, 79;610-621, 1956.
Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The metabolic basis of inherited disease. ed. 2, New York, McGraw-Hill, 1966.

SPRENGEL SYNDROME

Synonymy

S. of congenital elevation of the shoulder. S. of Sprengel deformity.

Syndromography

Clinic

This syndrome is characterized by presenting one scapula (rarely both) with vertical shortening and wider in its transverse diameter than the other. The scapula retracts during movement, except when attached to the dorsal spine or ribs. On the affected side the shoulder is elevated due to the shoulder malformation. There is frequently scoliosis that may be associated with torticollis.

Shoulder abduction above 90 degrees is impossible.

Paraclinical examinations

Chest and shoulder radiology. It is useful to establish the diagnosis exactly.

Syndromogenesis and etiology

The etiology is genetic and is transmitted by autosomal dominant inheritance.

Literature

Adams, R.D.; D. Denny-Brown and C. M. Pearson: Diseases of the muscle. ed. 2, Harper & Row, New York, 1962.
Sprengel, G.M.: “Die angeborene Verschiebung des Schulterblattes nach oben." Arch. f. klin. Chir. Berl., 42:545-549. 1891.
Wilson, M.G; V.G. Miksity and N.W. Shinno: Dominant inheritance of Sprengel's deformity." J. Pediat., 79:818-821, 1971.

STANESCU SYNDROME

Synonymy

Craniofacial dysostosis with diaphyseal hyperplasia.
Stanescu type osteosclerosis.

Syndromography

Clínic

The patients have a small skull, with depression of the frontoparietal and occipitoparietal sutures, poor jaw development and exophthalmos, short limbs, as well as short stature. Some cases also present defects of the spine and thorax with kyphoscoliosis and excavatum.

Paraclinical examinations

Radiology. Thin skull bones. Massively thickened short long bones. The increase in bone cortex occurs during or after puberty and increases with age.

Syndromogenesis and etiology

The etiology is genetic and an autosomal dominant inheritance is suggested; the basic defect is not known.

Literature

Dipierri. J.E and J.D. Guzmán: “A second family with autosomal dominant osteoclerosis-type Stanescu". Am. J. Med. Genet., 18:13-18.1984.

Maximilian, C. et al.: "Syndrome de dysostose cranio-faciale avec hyperplasie dyaphysaire." J. Genet. Hum., 29:129-139, 1981.

STEINBROCKER SYNDROME

Synonymy

S. of shoulder-hand. Reflex sympathetic dystrophy.

Syndromography

Clinic

It affects both sexes. Slight predominance in women. It is installed in patients over 50 years of age. Gradual stiffness, discomfort, weakness of shoulder and hand. Sudden, severe pain and stiffness. Edemae hyperesthesia of hand. It has 3 stages:

Stage I. Lasting 3-6 months. Complete commitment of hand and shoulder.

Stage II. Duration 3-6 months, partial or complete resolution of edema and vasospasm or vasodilation. Early trophic changes and contracture. muscle atrophy.

Stage II. Of variable duration. Atrophy and dystrophic changes. Finger contracture. Frozen shoulder.

Paraclinical examinations

Radiology of the chest, cervical area and shoulder.

Immunology. Rheumatoid arthritis should be excluded.

Syndromogenesis and etiology

It is considered a reflex dystrophy secondary to internal injuries: post-heart attack, herniated disc, post-trauma, post-hemiplegia, herpes zoster:. It has also been seen in tumors and without apparent cause (idiopathic).

Non-specific changes occur as can be seen in other types of capsulitis and periarthritis.

Literature

Hollander, J.L.: Arthritis and allied conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.
Steinbrocker, O.: "Painful homolateral disability of the shoulder and hand, with swelling and atrophy of the hand." Ann. Rheum. Dis., 6:80-84. 1947.

STILL SYNDROME

Synonymy

S. of acute juvenile rheumatoid arthritis. S. de Still- - Chauffard.

Syndromography

Clínic

More common in women. It sets in before puberty but never before 6 months of age.

Physical examination. High fever, erythematous rash. Pictures of pneumonitis, pericarditis, iridocyclitis, lymphadenopathy, splenomegaly, monoarticular or polyarticular arthritis.

Paraclinical examinations

Blood. Leukocytosis. Moderate anemia. Erythrose-accelerated sedimentation. LE cells: negative. Tests for rheumatoid arthritis positive in 20%.

Radiology. Demineralization. Space reduction articulate.

Syndromogenesis and etiology

Still's syndrome is related to collagenoses and autoimmune diseases.

A non-specific synovitis occurs, with granulation tissue and cartilage erosion. Nodal hyperplasia.

The etiology is unknown

Literature

Calabro, J.J.: Juvenile rheumatoid arthritis. In Hollander, J.L.: Arthritis and allied conditions, ed. 7, Philadelphia, Lea & Febiger, 1966.

Cornil, V.: "Mémoire sur les coincidences pathologiques du rhumatisme articulaire chroniques." Compt. yield Soc. of Biol., 4.s. 1(pt. 2):3-25, 1864-1865.

Still, GE: "On a form of chronic joint disease in children." Proc. Roy. M. & Chir. Soc. London, 9:10-15, 1896-1897.

SUDECK SYNDROME

Synonymy

S. of post-traumatic osteoporosis. S. of post-traumatic sympathetic dystrophy. S. de Sudeck-Kienbock. S. de Sudeck- -Leriche. S. of Kienböck. S. of Sudeck atrophy.

Syndromography

Clinic

Prevalence in the elderly and in women. Typical changes in the bones, however not always associated with trauma. There is pain, edema, stiffness, soft tissue atrophy, hyperhidrosis.

Paraclinical examinations

Radiology. Typical patterns of localized osteoporosis. Metaphyseal, subperiosteal and subarticular radiolucent bands.

Syndromogenesis and etiology

Various factors have been considered: trauma, infection, immobilization, burns, after a heart attack.

The etiology is unknown.

Literature

Allen, E.V.; N.W. Barker and E.A. Hines, Jr.: Peripheral vascular diseases. ed. 3, Philadelphia, Saunders, 1962.

Arnstein, A.R.: "Regional oste oporosis." Orthop. Clin. North Am., 3:585, 1972.

Guntheroth, W.G. et al.: "Posttraumatic sympathetic dystrophy. Dissociation of pain and vasomotor changes". Am. J. Dis. Child., 121:511, 1971.

Kienböck, R.: "Üeber akute Knochenatrophie bei entzündungs- prozessen an den Extremitäten (fäschlich sogenannte. Inaktivitätsatrophie des Knochens) und ihre Diagnose nach dem Röntgenbilde". Vienna. Med. Wschr. 51:1346-1348, 1901.

Sudeck, P.: "Ueber die acute enzundliche Knochenatrophie." Arch. Klin. Chi., 62:147-156, 1900.

Sudeck, P: “Ueber die acute enzundliche Knochenatrophie.” Verhan-dl. d. deutsch. Gesellsch. F. Chir., 29 (pt. 2):673-682, 1900.

CALCARE TENDINITIS SYNDROME

Synonymy

S. of the supraspinatus. S. of subdeltoid bursitis.

Syndromography

Clínica

This syndrome appears in sedentary workers, it prevails in men. It appears between 35-60 years of age, and is more common in the right shoulder, in a 2:1 ratio.

It is characterized by pain, sensitivity and limitation of shoulder movements, of varying intensity. Fever may appear in acute attacks.

Physical examination. The arm is voluntarily fixed in adduction, with impediment to abduction and rotation external.

Paraclinical examinations

Radiology. Calcium deposits in the tendon area affected.

Blood. Leukocytosis and accelerated erythrocyte sedimentation rate only in acute attacks.

Syndromogenesis and etiology

There is inflammation of one or more of the rotator tendons with calcium deposits around the tendon.

The etiology is unknown (post-trauma?).

Literature

Hollander, J.L.: Arthritis and allied conditions. Philadelphia, Lea & Febiger, 1966.

BICIPITAL TENOSYNOVITIS SYNDROME

Syndromography

Clínica

It is more common in women, the symptoms appear gradually or suddenly, especially at night. Pain of varying degrees appears with limitation to internal rotation and abduction. Tenderness of the anterior area of the humeral head.

Physical examination. Pain upon rotation maneuver and palpation of the affected tendon.

Paraclinical examinations

Radiology. It is difficult to pinpoint inflammation, but it allows other conditions to be ruled out.

Syndromogenesis and etiology

Trauma, change in temperature, humidity, idiopathic are suggested. Any of these factors can produce inflammatory changes in the bicipital tendon synovium.

Literature

Pasteur, E: Les aigiés de lépaule et la physiothérapie la teno-bursite bicipitale". J. Radiol. Electrol., 16:419-426, 1932.

TIETZE SYNDROME

Synonymy

S. of the costochondral junction. S. of tuberous chondropathy.

Syndromography

Clinic

Pain in one or more costal cartilages, increased by movement and coughing, radiating to the neck, shoulder and arm.

Physical examination. Edema of the upper costal cartilages. Sensitivity and slight hyperemia of the skin.

The second joint is the most frequently affected.

Paraclinical examinations

Electrocardiogram. To exclude cardiovascular pathology.

Chest radiology. Sometimes inflammation is seen in the affected areas.

Syndromogenesis and etiology

The symptoms are caused by a non-suppurative inflammation of the costal cartilages and concomital perichondritis.

tant.

The etiology is unknown.

Literature

Kayser, H.L.: Tietze's syndrome: a review of literature". Amer. J. Med., 21:982-989, 1956.

Tietze, A.: "Ueber eine eigenartge Haufung von Fallen mit Dystrophie der Rippenknorpel." Berl. Klin. Wschr., 58:829-831, 1921.

ACQUIRED TORTICOLIS SYNDROME0

Syndromography

Clinic

Painful contraction of the sternocleidomastoid muscle accompanied by tilting and deviation of the head.

Syndromogenesis and etiology

The etiology is attributed to: infections, neoplasms, traumatic or psychogenic.

Literature

Adams, R.D.: D. Denny-Brown and C.M. Pearson: Diseases of the muscle. ed. 2, Harper-Row, New York, 1962.

Lidge, R.T.; R.C. Bechtol and C.N. Lambert: "Congenital muscular torticollis; etiology and pathology." J. Bone Joint Surg., 39A:1165-1182, 1957.

Taylor, F.: "Induration of sternomastoid muscle." Trans. Path. Soc. London, 26:224-227, 1875.

SPASMODIC TORTICOLIS SYNDROME

Synonymy

S. of the crooked neck.

Syndromography

Clinic

It affects both sexes. The head is quickly pulled and appears twisted to one side. At the beginning there are light movements that gradually gain intensity. Emotional states and stress accentuate the symptoms. Over time the muscles may be permanently contracted and the head deviated to one side.

Physical examination. There is hypertrophy and contraction of the muscles, especially the sternocleidomastoid, the muscles of the posterior and lateral region of the neck and the upper part of the trapezius.

Syndromogenesis and etiology

It may be the sequel of chronic encephalitis, localized dystonia, or be part of the lenticular dystonia syndrome.

The etiology is unknown.

Literature

Poppen, J.L. and A. Martínez-Niochet: "Spasmodic torticollis." Clin. North America, 31:883-890, 1951.

MULTIFIDUS TRIANGLE SYNDROME

Syndromography

Clinic

It is characterized by acute and localized attacks of pain, isolated or more frequently recurrent, followed by localized discomfort below the posterior superior iliac spine.

It is most often triggered by exercise.

Physical examination. There is a point of sensitivity that increases under pressure and reproduces the typical irradiation.

Paraclinical examinations

Spine radiology.

Syndromogenesis and etiology

There is deep ligamentous or myofascial damage, exacerbated by trauma or inflammation that is located in the triangle of the multifidus muscle.

Literature

Bau wens, P. and A.B. Coyer: «The multifidus triangle' syndrome as a cause of recurrent low-back pain". Br. Med. J.. 2:1306-1307,1955.

Livingston, W.K.: "Back disabilities due to strain of multifidus muscle; cases treated by novocain injection." West J. Surg., 49:259-265, 1941.

CARPAL TUNNEL SYNDROME

Synonymy

S. compression of the median nerve in the carpal tunnel.

S. of carpal tunnel stenosing tenovaginitis.

Syndromography

Clinic

It predominates in women and is common during pregnancy and menopause. The patient reports paresthesias in the distribution of the median nerve; palmar aspect of the thumb, index, middle finger and radial half of the ring finger, weakness of the hands. Spontaneous pain in the forearm or shoulder and neck, the latter less frequently. It can be episodic or constant and is exacerbated by manual activity.

There is burning pain in the index, middle and ring fingers that often appears at night and wakes the patient up.

When symptoms are recent, sensory disturbance in the distribution of the median nerve may not be apparent. Sometimes there is weakness of the thumb.

Physical examination. Anesthesia of the thumb and fingers indicated above with atrophy of the thenar eminence, especially the abductor pollicis brevis; tenderness in the flexor tendons. The pain increases by flexing the wrist or with palmar flexion.

On percussion of the palmar aspect of the wrist, the tunnel sign or complete flexion of this area for more than a minute appears (Phalen maneuver). The symptoms of paresthesia or pain in the fingers are reproduced.

Paraclinical examinations

Electromyography. Sensory and motor conduction delay. Distal sensory conduction in the median nerve may appear before motor delay. Study of median nerve conduction showing late latency through the wrist.

Cervical spine x-ray. Necessary to rule out disc herniation at that level.

Syndromogenesis and etiology

This syndrome is due to compression of the median nerve between the palmar carpal ligament and the structures inside the carpus.

The most common cause is nonspecific synovitis that affects the flexor synovium in the carpal tunnel and compresses the nerve.
It can be observed in recent poorly consolidated fractures, tumors, and congenital anomalies.

It can also be a feature of systemic diseases such as rheumatoid arthritis, amyloidosis, acromegaly, myxedema, diabetes, gout.

Various causes are considered: tenosynovitis of the flexor tendons of the fingers, menstrual or pregnancy edema.

It is triggered by exercises that require wrist flexion, pronation, and supination (sewing, driving a car, operating computers), as well as trauma.

Literature

Marie, P. and C. Fois: “Atrophie isolée de léminence thénar dórigine néuritique, role du legament anulaire antérior du carpe dans la pathologénie de la lésion.” Rev. Neurol., 26:647-649, 1913.

Tobin, S.M.: "Carpal tunnel syndrome in pregnancy." Amer. J. Obstet. Gynec., 97:493-498, 1967.

TARSIUS TUNNEL SYNDROME

Syndromography

Clínica

Intermittent burning pain, paresthesias, cyanosis with numbness and coldness of the fingers occur after prolonged periods of standing or long walks; Sometimes these symptoms become more intense. The fingers and plantar regions are also affected. Many times the pain is relieved with massages and removing shoes.

Physical examination. Areas of hyposthesia and loss of discrimination of sensitivity in two points (positive tunnel sign) are seen. In advanced stages there is hypertrophy of the foot abductors.

Paraclinical examinations

Electromyography. It is the most accurate test.

Syndromogenesis and etiology

This syndrome can be produced by: post-traumatic fibrosis, hypertrophy of the abductor muscles of the great finger, tenosynovitis, or spontaneous entrapment. Any of these causes can result in chronic compression from the facial bands of the posterior tibial nerve deep along the medial border of the foot.

Literature

Edwards, W.G.; C.R. Lincoln; FH. III, et al.: "The tarsal tunnel syndrome, diagnosis and treatment." J.A.M.A., 207:716-720, 1969.

Keck, C: "The tarsal tunnel syndrome." J. Bone Joint Surg., 44A:180-182, 1962.

Pollock, I.J. and L. Davis: Peripheral nerve injuries. New York, Hoeber, 1933, pp. 32, 484-493.

NAIL-PATELLA SYNDROME

Synonymy

S. of hereditary osteo-onic-dysplasia. S. de Fong. S. de TourainеII. S. de Chatelain. S. of congenital iliac horns. S. of onychosteodysplasia. S. onychopatellar. S. of onion-hereditary osteodysplasia. S. of congenital iliac horns. S. nail-patella-elbow. S. de Fong.

Syndromography

Clinic

It is generally not discovered until the second or third decade. Both sexes are affected. It is almost always asymptomatic, in some cases patients report weakness and difficulty climbing stairs or dislocation of the kneecap.

Physical examination. Various nail abnormalities are observed (100% of cases): hypoplasia, longitudinal striae, thinning.

The most frequent changes are: hypoplasia, tarnishing and splitting of the nails.

Elbows: impossibility of complete extension, pronation or supination. Flexion is normal. Prominence of the medial epicondyle (90%).

Knees: kneecap usually smaller than normal or absent. Frequent dislocation and prominence of the medial femoral condyle with a decrease in the size of the lateral condyle (90% of patients).

The kneecap may appear divided into 3 parts, in a polygonal shape or be absent; Sometimes lateral dislocation of the patella occurs.

Pelvis: palpable, bilateral and symmetrical iliac horns (70%).

Eyes: iris pigmentation abnormalities (45%).

Occasionally there may be anomalies of the scapulae, lumbar lordosis, clinodactyly and arthritic changes of the joints.

These patients present nephropathy with a frequency of 30% of cases that can take the form of a nephrotic syndrome or glomerulonephritis, with isolated proteinuria and hematuria. Progressive renal failure is not common.

It is generally associated with other bone anomalies: sternum deformity, occult spina bifida, shoulder anomalies and others; as well as mental retardation, cataracts, etc.

Paraclinical examinations

Urine. Proteinuria in 40% of patients. Increase in mucoproteins.

bone radiology. Prominence of the medial epicondyle of the humerus, dysplasia with or without dislocation of the knee. Hypoplasia of the lateral condyle of the femur; hypoplasia or absence of the patella.
Horns on the iliac bone constitute a pathognomonic sign.

Kidney biopsy. Seen with the ordinary microscope, the lesions are nonspecific, but with the electron microscope a characteristic "moth bite" lesion is observed in the glomerular basement membrane, together with intramembranous collagen fibrils.

The prognosis is usually favorable if there is no kidney injury; if there is, death occurs in 8% of patients.

Syndromogenesis and etiology

It is of genetic etiology and is transmitted by autosomal dominant inheritance with variable expression, possibly linked to the loci that determines the ABO group.

The etiology is genetic.

Literature

Litle, E.M.: "Congenital absence or delayed development of patella." Lancet, 2:781-784, 1897.
Palacios, E.: "Hereditary osteo-onychodysplasia: the nail patella syndrome." Amer. J. Roentg., 101:842-850, 1967.
Simil, S. et al.: "Hereditary onycho-osteodysplasia (the nail-patella syndrome) with nephrosis-like renal disease in a newborn boy." Pediatrics, 46:61, 1970.
Turner, J.W.: "Hereditary arthrodysplasia associated with hereditary dystrophy of nails." J.A.M.A., 100:882-884, 1933.

VANBUCHEM SYNDROME

Synonymy

S. of generalized cortical hyperostosis. S. of endosteal hyperostosis. S. of late hyperphosphatasemia.

Syndromography

Clínic

This syndrome predominates in men. It is installed during puberty. It is generally asymptomatic for a long period of time, except when associated with mental deficiency.

It is characterized by osteosclerosis of the skull (base, vault), lower jaw, clavicles and ribs.

The progression of the disease can lead, through compression, to facial paralysis, optic atrophy and conductive deafness or perceptual hearing loss. Patients do not present such limitation in their physical activity.

Paraclinical examinations

Radiology. If a bone survey is performed, lesions are found in: skull, jaw, clavicle, ribs and the cortex of the diaphyses of the long bones. The flat bones are compact and absent of the diploe.

Blood. Elevated alkaline phosphatase.

Syndromogenesis and etiology

The basic defect is not known. The MIN code corresponds to autosomal recessive inheritance without an asterisk.

Literature

Turner, H.V. and D.H. Kelly, Jr.: "Steodysplasia with mental deficiency." J. Iowa Med. Soc., 58:260-268, 1968.
Van Buchem, F.S.P.: H. N. Hadders; R. Ubbens: "Uncommon familial systemic disease of skeleton: hyperostosis corticalis generalizata familaris." Acta Radiol., 44:109-120, 1955.

VOLKMANN SYNDROME

Synonymy

S. of post-traumatic muscle contraction. S. of ischemic contracture. S. of fibrous myositis.

Syndromography

Clínic

Burning pain, weakness or paresthesia in the hand and forearm.

Physical examination. Cyanosis, edema, coldness of the hands.

Fingers: flexion contracture.

Weak or absent radial pulse. Atrophy of the skin of the forearm and hand.

Paraclinical examinations

Radiology. Arteriography showing decreased blood flow.

Syndromogenesis and etiology

It is due to spastic or organic occlusion of arteries or veins of the arm caused by fracture or extrinsic compression. There is necrosis and degeneration of muscle fibers and ischemic degeneration of nerve fibers.

Literature

Boyes, J.H. (ed.): Bunnell's surgery of the hand. ed. 4, Philadelphia, Lippincott, 1964.
Volkmann, R.: “Die ischaemischen Muskellahmungen Kontracturen.” und Zentralbl. Chir., 8:801-803, 1881.

VON BECHTEREV-STRUMPELL SYNDROME

Synonymy

S. of spondylosis deformans. S. of rheumatoid spondylitis. S. de Bechterev. S. by Pierre Marie.

Syndromography

Clinic

Predominance in men. Insidious installation in the second or third decade. It is characterized by low back pain. Limited spinal flexibility, chest pain, kyphosis, scoliosis, back stiffness after a period of inactivity.

Physical examination. Loss of lumbar lordosis, reduction in spinal motility, eventual involvement of other joints, kyphosis and scoliosis.

Paraclinical examinations

Spine radiology.

Blood. Test for rheumatoid arthritis.

Syndromogenesis and etiology

Fibrosis of intervertebral capsules and ligaments that leads to ankylosis, with a chronic course and that produces complete rigidity of the spine in 10-15 years (bamboo cane). It may have lung involvement.

The etiology is unknown.

Literature

Bechterev, V.M.: "Oderevenelast' pozvonochikas iskrivleniemego, kak osobia forma zabolevaniia." Vrach. s. Peterburg, 13:899-903, 1892.

Epstein, B.S.: The Spine. Philadelphia, Lea & Febiger, 1962.
Marie, P.: Les rheumatismes deformants". Trib. Méd., 27:27-30, 1895.

Strumpell, A.: “Bemerkung uber dieg chronische Ankylosirende Entzundung der Wirbelsaule un der Ztschr. Huftgelenke Deutsche Nervenh., 11:338-342, 1897.

WHIPLASH SYNDROME

Syndromography

Clinic

Symptoms appear 1-2 days after a sudden extension of the neck, most often secondary to cervical trauma from a rear-end collision in a car. There is pain in the neck that radiates to both shoulders and arms.

Dysphagia, blurred vision, ringing in the ears, dizziness.

Paraclinical examinations

Radiology. The most common thing is the absence of injuries. A loss of cervical lordosis may occur initially and later an S-shaped defomity associated with vertebral alterations in the cervical spine.

In patients who present dysphagia at the beginning, retropharyngeal edema and/or hematoma may be

Literature

Coburn, D.E: “Vertebral artery involvement in cervical trauma.” Clin. Orthop. 24:61, 1962

MacNab, I.: "The Whiplash syndrome." Orthop. Clin. North. Am. 2:389, 1971.

RELATED MUSCULOSKELETAL SYNDROMES

Chondroectodermal syndromes

Ellis-Van Creveld.

Various forms of osteoauricular dysplasia:

a) True osteoauricular dysplasia.
b) Oto-vertebral.
c) Oto-palato-digital.
d) Oculo-oto-vertebral.
e) Ullrich-Feichtiger.
) Cranio-auriculo-radial.

Turner-Kieser.

McKusick.

Burke.

Lamy-Maroteaux.

Robinow-Silverman-Smith.

Pillay-Orth.

Tricho-rhino-phalangeal.

Trico-phalangeal.

Langer-Giedion.

Cranioectodermal dysplasia.

Maroteaux-Malamut.

Osteo-cutaneous.

Syndromes with metaphyseal dysplasia

Albers-Schönberg.

Cleidocranial dysostosis.

Multiple exostoses.

Hypophosphatasia.

Cartilage-hair hypoplasia.

Jansen.

Pycnodysostosis.

Pyle.

Schmidt.

Spahr.

Nail

Nail-patella.

Syndromes with hereditary bone dysplasia

Epiphyseal dysplasia

Blount-Barber.

Conradi.

Multiple epiphyseal dysplasia.

Spondyloepiphyseal dysplasia.

Physeal dysplasia

Achondroplasia.

Bone dysplasia-retinal detachment-deafness.

Metaphyseal dysostosis.

Klippel-Trenaunay-Weber.

Maffucci.

Marfan.

Ollier.

Metaphyseal dysplasia

Albers-Schönberg.

Cleidocranial dysostosis.

Multiple exostoses.

Hypophosphatasia.

Pycnodysostosis.

Pyle.

Nail-patella.

Diaphyseal dysplasia

Ellis-Van Creveld.

Engelmann.

Idiopathic osteoporosis.

Van Buchem.

Van der Hoeve.

Syndromes with intrinsic shoulder pain

Cold shoulder.

Calcareous tendonitis.

Bicipital tenosynovitis.

Musculotendinous sicuff of the shoulder.

Syndromes with joint hypermobility

Cretinism.

Down.

Ehler-Danlos.

Marfan.

Genetic trait, or cachectic state.

Van der Hoeve.

Metaphyseal dysplasia

Albers-Schönberg.

Cleidocranial dysostosis.

Multiple exostoses.

Hypophosphatasia.

Pycnodysostosis.

Pyle.

Nail-patella.

Diaphyseal dysplasia

Ellis-Van Creveld.

Engelmann.

Idiopathic osteoporosis.

Van Buchem.

Van der Hoeve.

Syndromes with dystonic myopathy

with myasthenia

Erb-Goldflam (pseudoparalytic myasthenia gravis).

Transient myasthenia of the newborn.

Lambert-Eaton.

Symptomatic myasthenia in the course of autoimmunopathies.

Symptomatic myasthenia in Basedow disease.

Symptomatic myasthenia with lupus erythematosus and palmoplantar keratosis.

MCArdle.

Denny-Brown.

With periodic paralysis

Westphal.

Gamstorp.

Normokalemic periodic paralysis (Poskanzer and Kerr).

Paramyotonic periodic paralysis (Becker).

Myotonic episodic adynamia.

Achor-Smith.

Bartter.

Albright-Hadorn.

Lightwood-Albright.

with myotonia

Thomsen (myotonia congenita).

Eulemburg (paramyotonia congenita).

Gamstorp-Wohlfahrt.

Neuromyotonia.

Schwartz-Jampel.

Rigid man.

Curschann-Batten-Steinert (myotonic dystrophy).

Rieger.

Iridodentaria (Weyers).

With myatonia

Oppenheim.

Prader-Wili.

Camurati-Engelmann.

Benign congenital essential hypotonia (Walton).

Minor-Oppenheim.

Mitochondrial myopathy syndromes

Ernster-Luft.

Megaconial myopathy.

Normokalemic periodic paralysis (with craving for salt).

Myopathic syndromes

Syndromes due to congenital defects skeletal muscles.

Incomplete regional achondroplasia (with dysplasia abdominal muscle).

Arthrogryposis.

Congenital defects of the pectoral muscles.

Deficiency of abdominal muscles.

Möbius.

Sprengel.

Stiff neck.

Syndromes with muscular dystrophy

Becker.

myotonic dystrophy.

Batten-Turner muscular dystrophy.

Duchenne muscular dystrophy.

Muscular dystrophy (Limb-girdle).

Leyden-Möbius type muscular dystrophy.

Congenital benign hypotonia.

De Lange type muscular dystrophy.

Progressive ophthalmoplegic dystrophy.

Duchenne in women.

Floppy infant.

Landouzy-Dèjerine.

Lewis-Besaut.

Distal Gower myopathy.

Hypertrophic myopathy.

Oculopharyngeal.

Oppenheim.

Neuromyopathic syndromes

Amyostatic.

Aran-Duchenne.

Congenital hereditary ataxia-cataract-dwarfism -mental deficiency.

Charcot-Marie-Tooth.

Coreiform.

Dejerine-Klumpke.

Dèjerine-Sottas.

Forerster.

Benign congenital hypotonia.

Rigid man.

Kugelberg-Welander.

Lenoble-Aubineau.

Progressive bulbar palsy.

Roussy-Cornill.

Roussy-Levy.

Tic.

Werdning-Hofmann.

Myositic syndromes

Fibrosis-myositis of the psoas.

Fibrositis.

Latissimus dorsi.

Anterior abdominal wall.

Anterior wall of the thorax.

Petges-Clejat.

Polymyalgia rheumatica.

Polymyositis.

Piriformis.

Straight right

Multifidus triangle.

Traumatic myopathic syndromes

Bywater.

Exertional thabdomyolysis.

Straight muscle.

Nielsen.

Overuse.

Shin splint.

Tibialis anterior.

Wattenberg.

Wolkmann.

Myopathic syndromes: toxic, metabolic, endocrine

Hereditary periodic adynamia.

Amyloidosis.

Glycogen storage.

Universal calcinosis.

Haff.

Hoffmann.

Kocker-Debré-Sémélaigne.

Myopathy in hyperparathyroidism.

Neuropathy in chronic uremia.

Periodic hypokalemic paralysis.

Normocalcemic periodic paralysis with craving salt (pleoconial myopathy).

Normocalcemic periodic paralysis without salt craving.

Periodic thyrotoxic paralysis.

Osteochondrodysplasia syndromes

Nievergelt.

Langer.

Ulnofibular.

Reeves (boomerang).

Wegmann (mesomielic dwarfism with dislocation of the head of the radius and tibia).

Mesomielic dwarfism with hypoplasia of the radius and of the tibia.

Campomelia.

Lery-Weill.

Madelung deformity.

Pillay-Orth.

Weismann-Netter.

Robinow-Silverman-Smith.

Syndromes with osteolysis

Essential osteolysis.

Family osteolysis.

Acro-osteolysis group.

Post-traumatic osteolysis.

Secondary or concomitant osteolysis:

a) Tabes dorsalis.

b) Leprosy.

c) Psoriasis.

d) Progressive scleroderma.

e) Alterations in arterial irrigation.

f) Diabetes mellitus with neuropathy.

g) Post-freezing.

h) Electrical aftershock.

i) Post-intoxication.

j) Bone neoplasia.

Syndromes with generalized osteolysis

I. Primary

Albers-Schöömberger:

a) Early malignant recessive hereditary osteosclerosis with bone fragility.

b) Benign dominant hereditary late osteosclerosis

na.

Pycnodysostosis.

Dysosteosclerosis.

Sclerosteosis.

Pyle.

Craniometaphyseal dysplasia.

II. Secondary

Osteosclerosis in poisoning of: lead, strontium or phosphorus.

Osteosclerosis as an epiphenomenon of fluorosis (Spira).

Osteosclerosis in hypercalcemia.

Osteosclerosis in renal tubular acidosis.

II. Reactive

Osteomyelosclerosis.

Syndromes with polymyositis

I. Primary

Primary chronic polymyositis with secondary muscular dystrophy.

Polymyositis and polymyalgia rheumatica.

Dermatopolymyositis (Wagner-Unverrich).

Dermatomyositis with poikiloderma (Petges-Clejat).

Acute polymyositis with myoglobinuria (Gunther II).

Focal myositis in Libman-Sacks (visceral lupus erythematosus)

Polymyositis in scleroderma (scleroderma- myositis).

Focal myositis in Kussmaul-Maier (periarteritis nodosa).

Focal myositis in Besnier-Boeck-Schaumann (sarcoidosis)

Ocular myositis:

a) Acute exophthalmic ocular myositis.

b) Oligosymptomatic chronic ocular myositis.

c) Ocular neuromyositis.

II. Secondary

Toxiinfectious myositis (toxoplasmosis, Coxsackie virus, acute epidemic myositis.

Parasitic myositis: trichinosis, protozoa (Chagas disease)

Concomitant myositis in metabolic diseases and enzymopathies (hyperuricemia, diabetes mellitus, lipothesaurosis, Teutschländer Erb-Goldflam and others).

Myositis concomitant with poisoning (narcosis, carbon monoxide poisoning and curare).

Concomitant myositis with neoplasms (metacarcinomatous neuromyopathy: Denny-Brown) with paraneoplasias.

Myositis in psoriasis vulgaris (psoriatic myopathy).

Overuse syndromes (OVERUSE)

Musculotendinous areas.

Shin splints.

Tibialis anterior.

Forearm sphicts.

Hamstring's origin.

Tennis player's elbow.

Golfer's elbow.

Player's elbow.

Javalinist's elbow.

Joints

Talotibial exostosis.

Chondromalacia of the patella.

Twist of the foot (sprain).

Soccer player's ankle.

Bones.

Stress fracture, march fracture (synonymous).

Muscles

Buekseb.

Exertional rhabdomyolysis.

Tendons

Achilles tendonitis.

Tendinitis (patellar).

Abductor tendonitis.

Trochanteric tendonitis.

Subcromial tendonitis.

Tendon Sheats.

Tenosynovitis.

Connective tissue syndromes associated with carcinomas

Rheumatoid arthritis.

Dermatomyositis.

Dysacromelias.

Fibrositis.

Lupus erythematosus.

Polymyositis.

ACROPARESTHESIAS SYNDROME

Synonymy

S. of nocturnal acroparesthesia. S. of Schultz acroparesthesia. S. of sleep acroparesthesia.

Syndromography

Clinic

More common in adult women, although it can be observed in both sexes. This syndrome is characterized by an abnormal sensation of numbness, tingling, burning, stabbing or burning pain, tension, compression or other unpleasant sensations in the hands, less frequently in the feet, which disappear with rubbing and pressure, allowing normal sensitivity to be restored. The patient has the sensation that the fingers and toes are asleep.

Atrophy of the abductor pollicis brevis and opponens pollicis muscles has been observed.

Syndromogenesis and etiology

This syndrome is due to compression of a nerve. It is seen in carpal tunnel syndrome, spinal cord tumors, herniated discs, and cervical rib syndrome.

It has also been observed in rheumatoid arthritis, myxedema, acromegaly and multiple myeloma.

Literature

Padilla, T.; P. Cossio: Semiology of the nervous system. 4th. ed., Buenos Aires, El Ateneo, 1944. p.321.

Grinker, R.R. and A.L. Sahs: Neurology. ed.6, Springfield, Thomas, 1966.

ADIE-CRITCHLEY SYNDROME

Synonymy

S. of Fulton. S. of forced grasping. S. of the premotor area.

Syndromography

Clinic

It is characterized by the fact that when an object is placed in the palm of the patient's hand, it

He grasps it perseveringly without letting go. A similar motor equivalent is produced when the sole of the foot is stimulated and the fan sign appears (response to the plantar reflex with the separation of the toes).

Syndromogenesis and etiology

It is due to a tumor of the contralateral superior frontal gyrus.

Adie and Critchley attribute a localizing value to the phenomenon of forced grasping, corresponding to Brodmann's area 6 on the opposite side. Fulton considers the sign of the fan as a locator in this area in which the reflex center of grasping would also reside.

Literature

Grinker, R.R. and A.L. Sahs: Neurology, ed. 6, Springfield, Thomas, 1966.

Walshe, FM.R.: "On the 'syndrome of the premotor cortex' (Fulton) and the definition of the terms premotor' and 'motor': with a consideration of Jackson's views on the cortical representation of movements." Brain, 58:49-80, 1935.

ADVERSE SYNDROME

Synonymy

S. of compulsive rotation. Adverse premotor epilepsy.

Syndromography

Clinic

Contralateral head rotation movements, ipsilateral miosis. Compulsive rotation when the patient tries to move forward. It may be accompanied by nystagmus.

Syndromogenesis and etiology

The adverse syndrome is consecutive to surgical ablation of the Brodmann area of the frontal lobe.

Literature

Eslinger, P.J. and A.R. Damasio: "Severe disturbance of higher cognition after bilateral frontal lobe ablation." Neurology, 36:1731, 1985.

BROCA'S APHASIA SYNDROME

Synonymy

S. of cortical motor aphasia. S. of total motor aphasia.

S. of expressive or executive aphasia.

Syndromography

Clinic

It is characterized by a loss of the motor component of speech and conservation of the sensory component. There is a verbal muteness with the inability to speak spontaneously, to repeat what has been heard,

and reading aloud. Sometimes the patient has few linguistic remains (short words, syllables or isolated phonemes) that are pronounced when he tries to express himself verbally. Emotional language is preserved. It is accompanied by agraphia (inability to write spontaneously or by dictation).

Syndromogenesis and etiology

The primary cause is a lesion of the posterior third of the inferior frontal gyrus (Broca's area) of the dominant hemisphere. Broca's area is adjacent to the lower motor cortex, which is why motor aphasia can be associated with a certain degree of paresis of the hand and right face and sometimes with orofacial apraxia, for which the patient is unable to wink, whistle or cough.

All disorders that affect the dominant hemisphere can cause aphasia: tumors, abscesses and hemorrhages, but the most common cause is an infarction of the middle cerebral artery territory.

Literature

Alexander, M.P.; M.A. Schmith: "The aphasia syndrome of stroke in the left anterior cerebral artery." Arch. Neurol., 37: 97, 1980.

Broca, P.: "Sur le siege de la faculté du language articulé, avec deux observations d'aphémie." Bull. Soc. Anat. Paris, 36:330- -357,1861.

Damasio, A.R.: "The neurological basis of language." Ann. Rev. Neuroc., 7:127, 1984.

Damasio, H. and A.R. Damasio: "The anatomical basis of conduction aphasia." Brain, 103:337, 1980.

WERNICKE'S APHASIA SYNDROME

Synonymy

S. of total sensory aphasia. S. of Pick-Wernicke-Bastion S. temporoparietal.

Syndromography

Clinic

Wernicke's aphasia encompasses a variety of syndromes produced by lesions of the posterior perisylvian structures or the posterior temporal, parietal and occipital regions supplied by the middle cerebral artery.

It is characterized by an alteration of the entire range of language behavior. When the lesion is in the temporal lobe, the main disorder is more evident in the language; On the contrary, when the parieto-occipital region is affected, the written word is the most affected.

This aphasia is characterized by being fluid, speaking and writing efforts for communication are affected, accompanied by an inability to visual and auditory comprehension (central aphasia). When reading, the patient can repeat aloud and copy, but when doing so he repeats the words heard with defective pronunciation as if they were an echo. However, he is usually extremely talkative, as if he had excessive verbal permeability (logorrhea) although he makes multiple mistakes (dysphasia), uses one word for another (verbal paraphasia) or changes syllables (syllabic paraphasia) or alters letters of the word (literal paraphasia); other times he repeats phonemes or syllables (perseverations).

When the paraphasia is very pronounced, they speak unintelligible jargon (jargonophasia). There are cases in which the patient is not aware of language defects (anosognosia). In the In severe cases, the patient mumbles a series of incomprehensible syllables and cannot repeat them out loud. Reading is impossible (alexia) or very difficult (dyslexia), when reading aloud confuses words, syllables or letters (paralexia). There are difficulties writing (dysgraphia). Sometimes it is accompanied by difficulty in understanding and reproducing music (amusia) and disturbance in mimicry and gesticulation (dysmimia). In the most serious cases, he makes illegible lines on a page trying to write. In its milder forms or later in the evolution of the disease, the speech resembles that of a tired or distracted person and the abnormality is only detected by language tests. There is no motor disorder, but the syndrome may be accompanied by hemianopia.

Paraclinical examinations

Computerized axial tomography and nuclear magnetic resonance. They are very useful to locate the injury and its extension.

Arteriography. It is not reliable because arterial occlusion due to a cerebral embolism hardly shows the embolus, because it has already disintegrated or has been carried to smaller branches.

Tests with radionuclides. Brain scanning with radioactive substances is only useful in extensive lesions.

Syndromogenesis and etiology

It is considered that any condition that, when acting on the brain, produces lesions or destroys the areas corresponding to the cortical centers of the language analyzers, can cause an aphasic syndrome.

Consequently, it can be caused by: transient cerebral ischemia, cerebral infarction due to occlusion of the left middle and posterior cerebral arteries, extensive hemorrhages, tumors or major trauma.

The lesions, for the most part, are located in the perisylvian region of the dominant cerebral hemisphere, left in right-handed individuals (right-handed individuals): only 10% are located in the right hemisphere and in those cases they affect the posterior third of the first temporal gyrus (superior temporal gyrus) and the neighboring region of the supramarginal fold and the transverse gyrus.

Literature

Damasio, A.R. and N. Geschwind: "The neural basis of language." Ann. Rev. Neurosc., 7:127, 1984.

Wernicke, K.: "Der aphasische symptomekomplex." Breslau, M. Cohn und Weigert, 1874.

SYNDROME OF AGENESIA OF THE CORPOUS CALLOSOUM

Syndromography

Clínic

Agenesis of the corpus callosum may be accompanied by prominent forehead and deep eyes (enophthalmos). Microcephaly has also been reported. This syndrome is sometimes accompanied by idiocy and epilepsy.

In general, patients present a serious intellectual deficit, especially in the ability to abstract and reduce the ability to concentrate. Alterations in coordination, bilateral.

Paraclinical examinations

Computerized axial tomography and ultrasound. They are essential to make the diagnosis.

Syndromogenesis and etiology

It is caused by a partial or total agenesis of the corpus callosum and constitutes a congenital malformation. It is almost always an autopsy finding that can coexist with other malformations. It is due to multiple genetic etiologies and is part of polymalformative syndromes; however, it has been reported in isolation with evidence of autosomal recessive inheritance.

The basic defect is unknown.

Literature

Alpers, B.J. and F.C. Grant: "The clinical syndrome of the corpus callosum." Arch. Neurol. & Psychiat., 25:67-86, 1931.

Shapira, Y. and T. Cohen: “Agenesis of the corpus callosum in two sisters." J. Med. Genet.. 10:266-269, 1973.

Young, I.D. et al.: "Agenesis of the corpus callosum and macrocephaly in siblings." Clin. Genet., 28:225-230, 1985.

ALEXANDER SYNDROME

Synonymy

S. of fibrinoid degeneration of astrocytes. Dysmyelogenic leukodystrophy with megalobarencephaly. Hyaline neuropathy.

Syndromography

Clinic

This syndrome prevails in males and begins in childhood. It evolves with idiocy and manifest mental retardation.

A pyramidal syndrome and, frequently, seizures are observed. The most characteristic is early and progressive megalocephaly.

Syndromogenesis and etiology

There is a congenital and familial leukodystrophy. It seems that the process is secondary to a disorder in the formation of myelin that occurs in the fetal period, which causes great destructive changes in the cerebral white matter, most intense in the frontal lobes.

It is of genetic etiology with autosomal recessive inheritance.

The basic defect is not known.

Literature

Alexander, W. S.: "Progressive fibrinoid degeneration of fibrillary astrocytes associated with mental retardation in a hydrocephalic infant." Brain, 72: 373-381, 1949.

ALPERS SYNDROME

Synonymy

Diffuse progressive degeneration of the cerebral gray matter. Infantile progressive cerebral poliodystrophy. S. de Chrinstensen-Krabbe.

Syndromography

Clinic

It is characterized by seizures, myoclonus, choreoathetosis. dementia and cerebellar ataxia.

Syndromogenesis and etiology

It is considered to be produced by anoxia as a consequence of trauma. These lesions cause degenerative changes in the middle layer of the cerebral gray matter that may also be present in the basal nuclei and the cerebellar cortex.

Literature

Alpers, B. J.: "Diffuse progressive degeneration of the gray matter of the cerebrum." Arch. Neurol. & Psychiat., 25:469-505, 1931.

INTERNAL CAROTID ARTERY ANEURYSM SYNDROME

Synonymy

S. of the foramen lacerum (anterior torn foramen or sphenoidal cleft).

Syndromography

Clinic

The rupture of an aneurysm in the internal carotid can cause a syndrome that, according to its location in the extracranial or extracavernous course of the artery, presents with three clinical varieties:

1. If it evolves towards the midline it produces symptoms of a pituitary tumor: homonymous bitemporal hemianopsia due to seizure of cranial nerve II (optic nerve); It rarely causes a clinical picture of anterior hypopituitarism.
2. When it grows forward, vision loss occurs due to seizure of cranial nerve II. Compressive manifestations are rare.
3. If it has a later development (most common variety), it determines involvement of cranial nerve III (oculomotor nerve).

In its intracavernosal course it rarely suffers rupture and when this occurs, it affects cranial nerves II, IV, VI and the ophthalmic branch of the trigeminal nerve.

Discrete exophthalmos is found due to disorders of the orbital venous circulation that drains into the cavernous sinus. Sometimes a murmur is perceived in the frontal region and especially in the eye.

There is unilateral paralysis of the extrinsic muscles of the eye, eyelid ptosis, divergent strabismus. Mydriasis and loss of pupillary light reflexes and accommodation, eye and/or facial pain (due to compression of the trigeminal ganglion); migraine-type headache.

The rupture of the aneurysm generally surprises the patient during physical exertion (defecation, coughing, intercourse). It is manifested by the appearance of a sudden headache with temporary loss of consciousness lasting a few minutes or seizures.

Symptoms depend on intracranial pressure, the presence or absence of hematoma, ischemia and acute hydrocephalus.

Subsequently, signs of meningeal irritation occur: neck stiffness, preretinal hemorrhages, papilloedema and jugular engorgement.

Paraclinical examinations

Skull radiology. Erosion of the sella turcica and, on occasion, enlargement of its bottom.

Computerized axial tomography. It is the method of choice, it demonstrates the presence, size and location of the hemorrhage.

Nuclear magnetic resonance. Much more sensitive to make the differential diagnosis.

Syndromogenesis and etiology

The degenerative lesion of the elastic tunica media gradually distends the arterial walls and leads to aneurysm formation.

It is due to congenital vascular malformations (the most common), degenerative vascular diseases: atherosclerosis. It can also be caused by vascular syphilis and infectious embolic arteritis as a complication of acute and subacute endocarditis (Streptococcus viridans).

Literature

Dailey, E.J.; J. A. Holloway; RE. Murto, et al.: "Evaluation of ocular signs and symptoms in cerebral aneurysm. Arch. Ophthal. (Chicago), 71:463-474, 1964.

Fisher, C. H. et al.: "Cerebral vasospasm with ruptured saccular aneurysm: The clinical manifestation." Neurosurg., 1:245, 1977.

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Mckissoch, W. et al.: "An analysis of the result of treatment of ruptured intracranial aneurysm. A report of 722 consecutive cases," J. Neurosurg.. 17: 762, 1960.

Toole, J E: Cerebrovascular disorders. Raven Press, New York, 1984.

Wilkins, R. H.: *Natural history of intracranial vascular malformation: A review." Neurosurg., 16:421, 1985.

CEREBELLARPONTON ANGLE SYNDROME

Synonymy

Tumor of the cerebellopontine angle. St. of Cushing II. S. heterolateral cerebellar.

Syndromography

Clínica

This syndrome almost always begins with vestibular or cochlear symptoms (seizure of the VIII nerve or vestibulocochlear nerve) that cause: dizziness, tinnitus, deafness, balance disorders and nystagmus. Progressive unilateral hearing loss and tinnitus.

There is abolition of the ipsilateral corneal reflex due to the seizure of the Vo trigeminal cranial nerve; At first it is only observed when the patient is in a lateral decubitus position, accompanied by painful paresthesias and sensitivity disorders in the trigeminal innervation territory. Deviation of the jaw towards the affected side when opening the mouth and peripheral facial paresis. These symptoms are caused by injury to the VII or facial nerve.

Eventually there may be seizure of the IX and X cranial nerves and sometimes the XI.

There are cerebellar symptoms (compression of the neighboring cerebellar hemisphere): dysmetria, tremor, ataxia (more frequently in the lower limbs), nystagmus and adiadochokinesia. Compression of the pons and medulla oblongata may produce contralateral pyramidal symptoms.

Symptoms of intracranial hypertension may be present, although "generally" late: headache and vomiting. There are characteristic pains located in the occiput and nape of the neck, more intense on the side of the injury.

Paraclinical examinations

Skull radiology. At the level of the petrous portion of the temporal bone, distention of the internal acoustic pore and decalcification of the tip of the rock can be seen in Stenvers and Towne positions, which allow comparison of the images of both rocks.

Audiometry. Alteration of brainstem auditory evoked potentials (there may be false positive results).

Computerized axial tomography. When the tumor is large extracanalicular. Magnetic nuclear resonance. Shows more accurate images.

Syndromogenesis and etiology

This syndrome is due to neoplastic lesions such as acoustic neuroma, the most common cause; and vascular disorders, less common.

Literature

Acers, T. E. and R. Tenney: "Ocular symptomatology of posterior fossa tumors." Amer. J. Ophthal., 65:872-876, 1968.

Graws, JE. et al.: "Cerebrovascular complications in patients with cancer." Medicine, 64:16, 1985.

Hort, A. et al.: "Acoustic tumors: Atypical features and recent diagnostic test." Neurology, 33:211, 1983.

Keschner, M. and M. Grossman: "Cerebellar symptomatology evaluation on the basis of intracerebellar and extracerebellar lesions." Arch. Neurol. Psychiat., 19:78-94, 1928.

ANTON-BABINSKI SYNDROME

Synonymy

S. of unilateral asomatognosia. S. of anosognosia.

Syndromography

Clínic

It is characterized by the patient who suffers from partial paralysis or hemiplegia, appearing indifferent or unconscious of the disorders they present, which can manifest in different ways, sometimes acting as if nothing matters to them, which includes a somatic sensory defect, as well as conceptual denial of the paralysis, negligence and even alteration of the visual perception of that hemibody (hemisomatognosia) and cerebral dullness. These disorders are usually associated with other abnormalities, for example: the patient appears gloomy, inattentive, disinterested and with varying degrees of general confusion with indifference to failure, or the feeling of having lost something, visual and tactile illusions when touching the affected area, hallucinations of movement and alloesthesia (stimulation of one side can be perceived by the opposite side).

It can be combined with homonymous hemianopia or lack of visual attention, neglect of a part of the body when dressing (apraxia), recognition of simultaneous and bilateral stimuli (sensory extension), deviation of the head and eyes towards the side of the injury, as well as twisting of the body in the same direction.

Syndromogenesis and etiology

This syndrome is due to lesions of the cerebral cortex and white matter of the right parietal lobe (non-dominant) that extend to a variable degree to the frontal motor areas and the temporal and occipital lobes. When the lesion is located in the left hemisphere, these disorders are rarely observed, in part because the presence of aphasia does not allow adequate exploration of other functions of the parietal lobe.

Literature

Zee, O. S. and R. J. Leigh: Disorders of ocular movement. FA. Davis Co., Philadelphia, 1983.

APRACTOGNOSIA SYNDROME

Synonymy

S. right parietooccipital. Dyspraxia. Use agnosia.

Syndromography

Clínic

It is convenient to clarify the meaning of apraxia (a: privative particle; praxis: action): Impossibility of performing acts (movements adopted towards an end) despite preserving motor integrity (without ataxia or paralysis) and psychic integrity (without mental or perceptual alteration), which prevents understanding the act or perceiving its elements.

Classification of apraxias (according to Liepman):

1. Motor apraxia. It can be ideokinetic and kinetic of the members.

In ideokinetic motor apraxia, motor engrams are preserved intact. The disorder resides between these and the other parts of the brain (interruption between the ideation and limb centers). There is an impossibility of performing complicated acts (tying a tie, lighting a cigarette). Sometimes there is perseveration (stereotypical performance of some elementary movement).

It is almost always localized (one limb, or an entire side). Kinetic motor apraxia of the limbs is produced by disorder or alteration of motor engrams. The individual has a precise and orderly mental representation of the different movements that make up the act to be performed; that is, the kinetic formula, but the members in charge of executing them cannot do it.

He cannot perform even the most basic movements. If he is ordered to pick up a glass of water, he will put his hand over it, but will not be able to grab it. It reminds us of the child who learns to perform a movement that is new to him for the first time.

2. Ideatory apraxia. The motor engrams remain unchanged, but the patient does not have the precise mental representation of the act to be performed. It is produced by intellectual disorders in the sphere of memory: attention, gnosia, association, etc. When told to light a cigarette, he either brings it to his lips and tries to smoke it without having lit it or tries to light the match with the cigarette. It seems like you have a lack of attention. It is central, affecting the entire body (utilization agnosia).

A variant of this type of apraxia is amnestic apraxia in which the patient cannot perform acts on his own initiative, but when done by imitation. Another variant is constructive apraxia, in which the patient can recognize but not reproduce the figures shown to him.

Syndromogenesis and etiology

The most frequent causes are: hemorrhages, thrombosis and embolisms in the anterior, middle, and posterior cerebral arteries and their branches. It is more common in the left cerebral hemisphere (in dextromaniacs) and in the parietal region (supramarginal gyrus and angular gyrus); lesions of the corpus callosum, apraxia of the face; at the level of the body, upper limbs, and at the level of the splenium of the corpus callosum, lower limbs.

Literature

Hecaen, H.; W. Pennfield:; C. Bertrand et al.: "The syndrome of apractognosia due to lesions the minor cerebral hemisphere." A.M. A. Arch. Neurol. Psychiat., 75:400-434, 1956.

BLACK WIDOW BITE ARACHNIDISM SYNDROME

Synonymy

S. of latrodectism due to black widow bite.

Syndromography

Clínica

After being bitten by the spider known as Black Widow, the patient presents diffuse, central and peripheral nervous excitability, which is manifested by convulsions and contractures, headache, hypertension and vasoconstriction. At the site of the bite, pain occurs and between 15-60 minutes, erythema is noticed around the tiny point of the bite, as well as cramps that gradually spread and can affect the limbs and trunk.

A 'board' abdomen is produced, but not painful on palpation, accompanied by nausea, vomiting, headache, sweating, hyperreflexia, contortions, tremor of hands and feet and, on occasions, systolic hypertension, with great agitation due to intense pain.

Vision and language disorders may occur, as well as respiratory distress.

Syndromogenesis and etiology

The syndrome is due to the bite of the female Black Widow spider (Latrodectus mactans or botonoid). The substance that causes the clinical picture is a neurotoxic protein from the venom inoculated by the spider, which binds to the presynaptic nerve endings and releases the transmitters contained therein. Post-synaptic depolarization causes muscle cramps in the affected limb that later become generalized.

Literature

Key, G F.: "A comparison of calcium gluconate and methocarbamal on the treatment of latrodectism. Am. J. Trop. Hyg., 30:273, 1981.

Kobernick, M.: "Black widow spider bite." Am. Farm. Physician, 29:241, 1984.

ARAN-DUCHENNE SYNDROME

Synonymy

S. of Aran-Duchenne atrophy. S. of myelopathic muscular atrophy. S. of Charcot amyotrophic lateral sclerosis.

Syndromography

Clínic

Symptoms predominate in men, in the fourth or sixth decade. There is symmetrical weakness in the small muscles of the hands, particularly the interossei, thenar eminence, and hypotena. Fibrillation and atrophy appear rapidly, gradually extending to the muscles of the arms, shoulders and trunk. There are generally fasciculations.

Paraclinical examinations

Any of them that is investigated gives negative results.
gatives.

Syndromogenesis and etiology

At least three autosomal recessive forms have been described:

1. Juvenile amyotrophic lateral sclerosis in which a DNA repair defect is suggested.
2. Amyotrophic lateral sclerosis concomitant with juvenile dementia.
3. Amyotrophic lateral sclerosis with polyglycan bodies.

Three autosomal forms have also been identified.
minants:

1. Amyotrophic lateral sclerosis with progressive and rapid loss of motor function. Pathological changes are limited to the cells of the anterior horn of the pyramidal tract.
2. Amyotrophic lateral sclerosis-Parkinson and dementia.
3. Amyotrophic lateral sclerosis with dementia.

Clinical heterogeneity suggests genetic and pathogenesis heterogeneity.

Literature

Arán, F. A.: "Recherches sur une maladie non encore decrit du systeme musculaire (atrophie musculaire pregressive)." Arch. Gen. Med. Paris, 24:172-214, 1850.

Duchenne, G. B.: *Etude comparee des lesions anatomique dans látrophie musculaire progressive et dans la paralysie generale." Union Med. Prat. Fr., 7:202, 1853.

Erbsloh, F.; K. Kunze; B. Recke et al.: "Die myatrophische Lateralsklerose, Klinische, elektromyographische und biopticsch-histologische Untersuchungen an 112 Kranken." Deutsch. Med. Wschr., 83:1131-1141, 1968.

ARNOLD-CHIARI SYNDROME

Synonymy

Anold-Chiari malformation. S. of basilar impression. S. by Arnold-Chiari-Celand.

Syndromography

Clínic

This syndrome affects both sexes, mainly affecting children who have spina bifida, especially lumbar.

Although Chiari divided it into four types, it has currently been restricted to Chiari types I and II, that is, the bulbocerebellar malformation without myelomeningocele (type I) and with myelomeningocele (type II). Type II is an occipital meningoencephalocele and type IV (doubtful) is a cerebellar

In type I (without meningocele), neurological symptoms may appear in adolescence or towards the third or fourth decade of life. Symptoms may be caused by increased intracranial pressure, progressive cerebellar ataxia or syringomyelia; or the patient may present disorders of the cerebellar type and the last cranial nerves (medulla and spinal cord) and headache. In 25% of cases it has a short neck or "bull's neck".

In type II (with myelomeningocele) the symptoms are produced by hydrocephalus due to the compression of the ectopic structures on the cerebrospinal fluid circulation pathways, thus we have: cervico-occipital headache, vomiting, visual disorders (diplopia), involvement of the long motor pathways (paralysis of the limbs), ataxia, seizure of the last cranial nerves (laryngeal stridor, tongue fasciculations, paralysis ster-nocleidomastoid, facial weakness, deafness).

Paraclinical examinations

Opaque myelography. It shows characteristic radiological images that corroborate the diagnosis.

Angiography. Reveals displacement of the posterior inferior cerebellar arteries.

Cerebrospinal fluid. Increased pressure and proteins in some cases.

Syndromogenesis and etiology

It is caused by congenital anomalies, possibly multifactorial, of the base of the brain, among the most frequent
are:

1. Extension of a flap of cerebellar tissue posterior to the medulla oblongata and the spinal cord within the cervical canal.
2. Displacement of the medulla oblongata and part of the fourth ventricle into the cervical canal.

This syndrome may also be accompanied by other alterations in the morphology of the central nervous system such as: elongation of the pons and medulla oblongata and narrowing of the Sylvian aqueduct; occlusion of the foramen occipital, occlusion of the cisterna magna, and the foramina of Luschka and Magendie open to the cervical canal.

Abnormalities of brain development (polymicrogyria) and an extension of the spinal cord to the sacrum, bone malformations may coexist: wide and posteriorly projected foramen occipital, flattening of the skull base (platybasia) or an underdeveloped foramen occipitalis. It is almost always accompanied by myelomeningocele and hydromyelia of the cervical cord is common.

Literature

Arnold, J.: "Myelocyste, Transposition von Gewecbskeimen und Sympodic." Beitr. z. Path. Anat. u.z. allg. Path., 16:1-28, 1894.

Chiari, H.: “Ueber Veranderlngen des Kleinhirns inflge von Hydocephalie des Grosshirns.” Deutsche Med. Wschr., 17:1172- -1175, 1891.

Vinken, D. J.: "Congenital malformations of the spine and spinal cord." Handbook of Clinical Neurology, vol. 32, American Elsevier Publ. Co., New York, 1978.

CAROTID ARTERY SYNDROME REDUNDANT

Synonymy

S. of intermittent obstruction of the internal carotid. S. of transient hemiparesis.

Syndromography

Clínic

Vertigo, transient hemiparesis or loss of vision in one eye when turning the head to one side, which disappears when the head is returned to the middle position.

Physical examination. The above symptoms are checked.

Paraclinical examinations

Carotid Doppler.

Arteriography.

Syndromogenesis and etiology

It is caused by a tortuosity of the carotid. When you turn the head, the glass twists and becomes clogged.

The cause is a possible congenital malformation or acquired dilation and tortuosity of the artery.

Literature

Kellogg. D. R. and L. L. Smith: Recurrent monocular blindness due to a redundant carotid artery." Arch. Surg., 95:908-910, 1967.

Quattelbaum, J.K.; E.T. Upson and R. L. Neville: “Stroke associated with elongation and kinking of the internal carotid artery: report of three cases treated by segmental resection of the carotid artery." Ann. Surg., 150:824-832, 1959.

Riser, M.; J. Geraud; J. Ducoudray et al.: "Dolicho-carotide interne avec syndrome vertigineux." Rev. Neurol., 85:145-147, 1951.

CAROTID ARTERY SYNDROME

Syndromography

Clinic

This syndrome can cause two clinical conditions depending on the intensity of the lesion: stenosis or occlusion.

If stenosis occurs, transient alterations are common, the most characteristic being the loss of ipsilateral monocular vision. Episodes of sensory disorders and paresis of the upper and lower limbs or the face on the opposite side are common, which can sometimes lead to hemiplegia and hemisensory disorders, which return to normal in minutes, hours or days. This condition may be accompanied by dysarthria when the dominant hemisphere is involved.

The occlusion can occur asytomatically or be preceded by transient manifestations that culminate in a cerebral attack. 50% of patients do not present previous manifestations.

The motor or sensory cortex may be affected, causing motor and sensory disorders of the contralateral hemibody, including the face. If the dominant hemisphere is involved, it is accompanied by language disorders. one third of patients present mild or severe ipsilateral or sometimes contralateral frontal or orbital pain. Approximately 15% develop Horner syndrome.

Physical examination. Strong palpation of the carotid artery in the neck is not advisable due to the possibility of dislodgement of an intra-arterial clot. Light palpation confirms loss of pulse, which can be complemented by the absence of pulse in the facial and superficial temporal arteries, when the occlusion occurs at the level of the bifurcation of the common carotid artery.

In stenosis of the internal carotid artery, a carotid murmur can be heard at the level of the bifurcation, which is located by placing the stethoscope at the level of the upper edge of the thyroid cartilage. In severe stenosis of the internal carotid, as it passes through the cavernous sinus, it is possible to hear a murmur at the level of the orbit that can also appear when the contralateral internal carotid is occluded, in this case due to compensatory collateral arterial circulation.

Diastolic retinal pressure on one side (determined by ophthalmic dynamometry) of less than 20 mm of mercury usually means that the ipsilateral common or internal carotid arteries are occluded. The presence of shiny or dull retinal emboli is another sign of carotid disease.

All neurological signs and symptoms of internal carotid occlusion may be present in common carotid occlusion. Both common carotid arteries may be occluded, causing aortic arch syndrome or pulseless disease (see Takayasu syndrome), characterized by: absence of carotid and radial pulses, dizziness upon lifting movements, recurrent loss of consciousness, headache, neck pain, transient unilateral or bilateral blindness, cataracts, retinal atrophy and pigmentation, or claudication of the chewing muscles.

Paraclinical examinations

Doppler.

Carotid angiography.

Intravenous digital subtraction angiography. Use computing and avoid the risks of arteriography.

Syndromogenesis and etiology

The syndrome is mainly caused by atherothrombotic lesions in the arterial wall of the common carotid.

Literature

Brice, J.C.; DJ. Dowsett and R.D. Lowe: "Haemo-dynamic effect of carotid artery stenosis." Br. Med. J.. 2:1363. 1964.

ANTERIOR CEREBRAL ARTERY SYNDROME

Syndromography

Clínic

The most common clinical picture caused by the anterior cerebral artery is characterized by decreased muscle strength and loss of sensitivity of the contralateral lower limb. The upper limb may be involved to a lesser extent, and more rarely the face. They are frequently associated with bladder sphincter disorders, mental confusion, contralateral grasping and sucking reflexes, and on some occasions, other symptoms such as: slowness and lack of spontaneity, laconic responses, distraction, and whispering.

Syndromogenesis and etiology

When the occlusion of the middle cerebral artery is proximal, it does not present symptoms, since the anterior communicating artery provides the necessary blood flow from the anterior cerebral artery on the opposite side.

When both arteries originate from the same anterior brain stem, the condition is more severe and both hemispheres are affected, causing paraplegia, incontinence of frontal origin and profound mental symptoms.

Literature

Mokry, D.B.; T.M. Sundt; O.N. Houser, et al.: "Spontaneous dissection of the cervical internal carotid artery." Ann. Neurol., 19:126, 1986.

MIDDLE CEREBRAL ARTERY SYNDROME

Syndromography

Clinic

When there is an obstruction of the middle cerebral artery, a clinical picture characterized by hemiplegia and contralateral hemianesthesia and homonymous hemianopia arises.

If the dominant hemisphere is affected, it may be accompanied by aphasia, and amorphosynthesis if the non-dominant hemisphere is affected. In lesions on the left side there is initially a global aphasia, predominantly motor with improvement in the understanding of the spoken and written word, accompanied by hesitant, dysmelodic and grammatically simplified language.
Lesions in the posterior parietal region can cause inferior quadrantonopia.

Syndromogenesis and etiology

The cortical distribution of the middle cerebral artery covers motor and somatosensory areas in relation to the upper limbs and the face and more sparsely with the lower limbs (lower two thirds of the motor and sensory homunculus) and, in addition, motor areas 4 and 6, the centers for lateral gaze, the language motor area or Broca's (dominant hemisphere), angular and supramarginal gyrus, upper part of the temporal lobe and insula, as well as the posterior arm of the internal capsule. Therefore, the lack of irrigation of these areas gives rise to the clinical picture.

The etiology in most cases is embolism, more rarely thrombotic lesions.

Literature

Wishnant, J. P. and D. D. Wiebers: Clinical epidemiology of transient cerebral ischemic attacks (TIA) in the anterior and posterior cerebral circulation. In Sundt, T.M. (ed). Occlusive cerebral vascular disease. Diagnosis and surgical management. P.A., W.B., Saunders Company, Philadelphia, 1987.

ANTERIOR CHORODAL ARTERY SYNDROME

Synonymy

S. de Von Monakow.

Syndromography

Clinic

Hemiplegia, hemianesthesia, hemianopsia contralateral to the injury. The clinical pattern is sometimes variable and hemianesthesia and hemianopsia may be partial.

Paraclinical examinations

Lumbar puncture.

Angiography.

Computerized axial tomography.

Syndromogenesis and etiology

The syndrome is caused by rupture or thrombosis of the anterior choroidal artery, caused by an aneurysm or tumor.

Literature

Kolinsko, A.: Ueber die Bezichung der Arteria choroidea anterior zum hinteren Schenkel der inneren Kapsul des Genhirnes. Vienna, 1891.

Steegmann, A.T. and D. J. Roberts: "The syndrome of the anterior chroidal artery." JA.M.A., 104:1695-1697, 1935.

FAMILIAR HYPOGONADIC CEREBELLAR ATAXIA SYNDROME

Syndromography

Clinic

Association of cerebellar syndrome (Marie's ataxia).

Friedreich's S., or neocerebellar S. with primary hypogonadotrophic hypogonadism.

Paraclinical examinations

Low gonadotropin excretion and increased steroid excretion.

Syndromogenesis and etiology

There is a total or partial failure of the pituitary gland to stimulate gonadal development at puberty, of unknown etiology.

Literature

Holmes, G.: "A form of familial degeneration of the cerebellum." Brain, 30: 466-488, 1907.

POST-INFECTIOUS CEREBELLAR ATAXIA SYNDROME

Syndromography

Clinic

It almost always appears in second childhood. It begins suddenly in healthy children or is preceded by a viral infection (measles, chickenpox or scarlet fever) as a rare complication, from the third to eighth day after the onset of the infection. Its clinical picture is very variable, the most common being acute seasonal ataxia and nystagmus during walking. severed language, intentional tremor.

Syndromogenesis and etiology

This syndrome is associated with viral infections (Coxsackie viruses A4, A7, A9, B3 and B4 and with ECHO viruses 6, 9 and 16.

Literature

Goldwyn, A. and A. M. Waldman: “Acute cerebellar ataxia in children: a report of three cases." J. Pediat., 42:75-79, 1953.

Klingman, W. O. and R. G. Hodges: "Acute ataxia of unknown origin children." J. Pediat., 24:536-543, 1944.

MARIE'S ATAXIA SYNDROME

Synonymy

S. de Pierre-Marie. S. of hereditary Menzel ataxia. S. of cerebellar heredoataxia.

Syndromography

Clinic

It occurs in both sexes after the age of 20. It is characterized by gait ataxia (cerebellar-spastic gait), a mixture of cerebellar hesitancy and pyramidal spasticity (in the standing position, the patient separates the legs and swings).

Spasticity in extension of the limbs and to a lesser extent in flexion. Sometimes clubfoot. The patient complains of cramps and painful attacks, but sensory disorders are rare. There is dysarthria, the word is slow, spasmodic with disorder in pronunciation due to an intentional perioral spasm; dysphagia. The reflections are preserved.

It may be accompanied by ophthalmoplegia and atrophy of the optic nerve, frequent nystagmus and sometimes Argyll-Robertson sign. Sometimes epileptic seizures appear and sometimes a certain degree of mental deficiency.

Syndromogenesis and etiology

It is a hereditary disorder that is usually dominant but sometimes recessive. There is cerebellar atrophy that mainly affects the cerebellar peduncles.

middle, pontine and olivary nuclei and to a lesser extent the dentate nuclei and the superior cerebellar peduncles, there are also lesions of the dorsal cardons of the spinocerebellar and corticospinal tracts.

Literature

Dow, R. S. and G. Moruzzi: The physiology and pathology of the cerebellum. University of Minnesota Press, Minneapolis, 1958.

Harding, A. E.: The hereditary ataxias and related disorders. Churchill Livingstone, Edinburgh, 1984.

Marie, P.: "Sur l'hérédo-atasie cerebelleuse." Sem. Med., 13:444-447, 1893.

ATAXIA-TELANGIECTASIA SYNDROME

Syndromography

Clinic

Early cerebellar type ataxia. Slow and scanty voice, there are no alterations in reflexes or sensitivity.

Oculocutaneous telangiectasia in the mucous membranes that begins in the eyelid region. Recurrent lung infections.

Paraclinical examinations

Pneumoencephalography. Signs of cerebellar atrophy.

Magnetic nuclear resonance.

Immunological study. Decreased IgA, as well as IgE and IgG. IgM may be increased.

Syndromogenesis and etiology

It is due to cerebellar degeneration with pathological alteration of Purkinje cells and atrophy of the white matter.

It is transmitted by autosomal recessive inheritance and is accompanied by pluriglandular involvement, variably decreased pituitary reserve, saccharine diabetes, adrenal corticosteroid hypoplasia and dysgenic ovaries.

Synonymy

EMBOLISM SYNDROME OF THE POSTERIOR CEREBRAL ARTERY

Syndromography

Clínica

Infarction caused by obstruction of the proximal branch of the posterior cerebral artery includes thalamic syndrome and mesencephalic syndromes.

It is characterized by hemiballismus or hemicoreoathetosis, vertical tremor, upward and outward deviation of gaze, and retraction nystagmus.

Lesions of the cortical branches frequently cause homonymous hemianopia, dyslexia, and a variety of visual hallucinations. Bilateral lesions cause cortical blindness.

When leptomeningeal collateral arteries from the middle and anterior cerebral arteries supply the posterior pole of the cerebral hemisphere, central vision may be preserved.

Bilateral occlusion of the posterior cerebral arteries can cause amnesia syndromes (Korsakof's amnesic state) by producing anoxia of the hippocampal formation of the temporal lobes or a paramedian thalamic infarction syndrome when the thalamoperforating branches are occluded.

Syndromogenesis and etiology

Atherothrombosis is due to occlusion of the vascular lumen by thrombi or emboli.

The lesions that produce this condition are: atherosclerosis, atherosclerotic lesions, atrial fibrillation and other arrhythmias, myocardial infarction with mural thrombi, bacterial endocarditis, complication of cardiac surgery, valve prostheses, endocardial vegetations, atherosclerosis of the aorta and carotid arteries, thrombosis of the cerebral arteries, thrombi in the pulmonary veins, complication of neck surgery and thorax, thrombophlebitis, arteritis, hematological disorders, carotid trauma, dissecting aortic aneurysm, arterial hypertension, complications of arteriography, hypoxia, etc.

Paraclinical examinations

The most commonly used tests in the study of cerebrovascular lesions are: Ophthalmodynamometry, oculoplethysmography and supraorbital directional Doppler. They are used to indirectly assess the pressure in the internal carotid artery.

They are abnormal when the luminal residual diameter of the atheromatous lesion is less than 1 mm and the distal pressure is greatly reduced.

Ultrasound. Reliably identifies atheromatous lesions at the common carotid bifurcation.

Quantitative spectral phonangiography. It calculates the residual diameter of the lumen of the internal carotid artery and allows us to differentiate between a murmur that originates in the carotid bifurcation and one that radiates from the base of the heart.
This test tends to be more accurate when the residual luminal diameter is 0. 9 - 2.5 mm.

Cerebral angiography (transfemoral catheterization and selective extracranial injection). It continues to be the most reliable method to assess the cerebrovascular tree. It can detect ulcerative lesions, severe stenosis and formation of mural thrombi at the level of the carotid bifurcation, siphon and intracranial vessels and collateral circulation around the arterial circle of the brain. It indirectly indicates decreased flow in the internal carotid system.

Intravenous digital subtraction angiography. It shows carotid occlusion or severe stenosis but does not always specify the degree of stenosis.

Computerized axial tomography. It allows calculating the degree and location of supratentorial cerebral impact, including small infarcts of the brain stem (vertebrobasilar).

Positron emission tomography. It allows qualitative and quantitative tomographic assessment of cerebral flow.

Nuclear magnetic resonance. It specifies the degree and location of the impacted tissue a few hours after the disorder.

LATE CEREBELLAR CORTICAL ATROPHY SYNDROME

Synonymy

S. of lamellar atrophy of Purkinje cells. S. of cerebellar corticoparenchymal atrophy. Cerebellar-olivary atrophy. St. of Alajouanine-Foix-Marie. S. of cerebellar cortical degeneration.

Syndromography

Clinic

It occurs in both sexXoS and characteristically in the sixth decade. It is characterized by the insidious progression of ataxia that begins in the lower limbs, resulting in abnormal posture and gait instability. Later, language disorders appear and the arms become ataxic. There may be nystagmus.

Paraclinical examinations

Computerized axial tomography. In advanced cases it shows signs of atrophy of the cerebellar cortex.

Syndromogenesis and etiology

In most cases it is inherited as an autosomal dominant trait. Degenerative changes occur in the superior vermis and adjacent parts of the cerebellar cortex (loss mainly of Purkinje cells).

Literature

André-Thomas: "Atrophy of Purkinje cells." Rev. Neurol., 13:917-924, 1905.

OLIVOPONTOCEREBELLAR ATROPHY SYNDROME

Synonymy

S. de Déjerine-André-Thomas.

Syndromography

Clínica

It occurs in both sexes in middle age or old age (after age 50), almost always slowly and insidiously.
It is characterized by the patient increasing the base of support during walking and the standing station hesitates, the trunk oscillates.

He has difficulty walking, hand tremors, and motor disorders for fine movements.
Physical examination. Muscle hypotonia. Except for the patellar, all other reflexes are absent. There is an absence of sensory changes.

Romberg's sign is negative. There are dysmetric movements of the lower limbs and slight intentional tremor in the upper limbs, irregular writing, slow spelled speech and slight nystagmus, mental deterioration.

Syndromogenesis and etiology

Of genetic etiology, it is inherited with an autosomal dominant character. There is degeneration of the middle cerebellar peduncles, the cerebellar white matter, and the pontine, olivary, and arcuate nuclei.

Literature

Déjerine, J. et A. Thomas: "L'atrophie olivo-ponto-cerébelleuse." Nouv. Iconog. Salpet., 13:330-370, 1900.

AVELLIS SYNDROME

Synonymy

S. of the spinothalamic tract. S. of the ambiguous nucleus. Ambiguouspinothalamic palsy. S. de Avellis-Longhis.

Syndromography

Clinic

It is characterized by dysphagia, unilateral loss of sensitivity to pain and temperature in the trunk and limbs.
It may be associated with Horner syndrome. Paralysis of the soft palate and vocal cords contralateral to the loss of sensation. There is contralateral hemiplegia.

Syndromogenesis and etiology

This syndrome is caused by lesions that affect the nucleus ambiguus of the X (vagus) and

The etiology can be: vascular, inflammatory or neoplastic.

Literature

Avellis, G.: "Klinische Beitrage zur halbseitigen Kchlkopflahmung." Berl. Klin., 40:1-26, 1891.
Fox, S.L. and G. B. West Jr.: "Syndrome of Avellis; review of literature and report of one case." Arch. Otolaryng., 46:773-778, 1947.

BABINSKI SYNDROME

Synonymy

Segmental spinal paralysis.

Syndromography

Clínica

The clinical picture observed is characterized by cerebellar hemiataxia, contralateral hemiparesis and loss of sensation in the trunk and limbs. Nystagmus and ipsilateral analgesia may occasionally be found on the face; other times, adiadochokinesia, lateropulsion and dysmetria.

It is also possible to observe associated Horner syndrome (enophthalmia, ptosis of the upper eyelid, miosis, increased tear secretion with absence of sweating on the ipsilateral side of the face and neck, facial hemiatrophy and ocular hypotonia).

Syndromogenesis and etiology

In cases of bulbar location, the clinical picture is produced by injury to the restiform body, Deiters nucleus and sympathetic fibers.

S. by Bailey Cushing

BALINT SYNDROME

Synonymy

S. of psychic paralysis of visual fixation. S. of cortical paralysis of visual fixation.

Syndromography

Clinic

Balint syndrome is characterized by the patient's inability to look ahead at a point that is in his peripheral visual field, being unable to move his eyes to follow a moving object and to estimate the distance between two objects located at different distances from him.

There is optic ataxia, which consists of the individual's inability to make voluntary movements in response to visual stimuli. The patient tries to catch an object by extending his hand in the wrong direction and manages to reach the object after repeated attempts. It also presents attention disorders (normal for all non-visual stimuli). When walking he bumps into any obstacle. He does not know how to orient himself although he knows and describes the route. The symptoms are bilateral but more marked on one side; they may be accompanied by language difficulties (agraphia and ideomotor apraxia).
There are tonic and motor disorders of the limbs and loss of coordination on both sides of the body. Stereoscopic vision and visual memory are preserved.

Paraclinical examinations

Radiology and computerized axial tomography. They show diffuse cortical atrophy.
Ophthalmological examination.
Electroencephalogram.

Syndromogenesis and etiology

It is due to destructive lesions of the occipital lobe in Brodmann's areas 18 and 19 of the dominant hemisphere.

Literature

Adams, R. P. and M. Victor: Principles of neurology. McGraw-Hill, New York, 1985.
Balint, R.: "Seelenlahmung des Schauens, optische Ataxia raumliche Storung der Aufmerksamkeit." Mschr. Psychist. Neurol., 25:51-81, 1909.

Damasio, A. R.: "Disorders of complex visual processing." In M. M. Mesulam (ed.) Principles of behavioral neurology. F. A. Davis Company, Philadelphia, 1985. pp: 259.

BALO SYNDROME

Synonymy

S. of concentric periaxial encephalitis. Balo disease.
S. of concentric unilobar sclerosis of Balo.

Syndromnography

Clinic

Balo syndrome is probably a histological variety of Schilder's diffuse periaxial sclerosis. It begins and evolves acutely, in both sexes, and generally develops from childhood. It presents with progressive spastic hemiplegia, paresthesias, aphasia and frequent signs of intracranial hypertension.

Syndromogenesis and etiology

In this syndrome, alternative bands of destruction and conservation of mnielin occur, arranged in the form of concentric rings. Apparently it is due to the centrifugal diffusion of some factor that attacks myelin.
The etiology is unknown.

Literature

Balo, J.: "Encephalitis periaxialis concentrica." Arch. Neurol. Psychiat., 19:242-264. 1928.

BARDET-BIEDL SYNDROME

Syndromography

Clinic

This syndrome is characterized by mental retardation, pigmentary retinopathy, obesity and hypogenitalism.

Of 32 patients examined, 100% had severe retinal dystrophy (only 6% had typical retinitis pigmentosa), 96% had obesity, and 40% had mental retardation. Hypogenitalism in men was 87.5% and menstrual irregularities were evident in 100% of women. All patients had syndactyly and/or brachydactyly, but polydactyly was present in only 56% of the cases. In addition, 45% had diabetes mellitus and 100% had abnormalities of kidney structure or function. In other patients there were cardiovascular anomalies.

Retinal pigment degeneration begins to manifest itself in childhood through twilight blindness (visual difficulty at dusk or in poorly lit rooms).

Examination of the visual field demonstrates the existence of an annular scotoma corresponding to the lesions located in the equatorial region of the retina. The condition worsens progressively and slowly in the following years of the patient's life. Due to the extension of the aforementioned annular scotoma, it reaches the periphery of the nasal visual field, leaving only a central area of vision and a temporal island that ends up being lost. Visual acuity is remarkably preserved even in very advanced stages, but then the patient cannot take care of himself, since with such a small field of vision he cannot orient himself sufficiently, for example, to cross a street. Finally, central vision decreases and the process sometimes stops around 40 years of age. There are cases in which vision ends up being lost, as the small remainder of the visual field that remains disappears.

It is also accompanied by deaf-muteness, idiocy, infantile adiposis.

Paraclinical examinations

Electroretinogram. To study the alterations of the retina.

Campimetry.

Renal ultrasound and intravenous pyelography. They allow kidney abnormalities to be identified (calyceal cysts, diverticula, anomalies of the calyces).

Dermatoglyphs. Hand shape abnormalities are checked.

Karyotype. To exclude chromosomal abnormalities.

Endocrinometabolic studies. They detect diabetes mellitus, hypogonadotropism, primary gonadal defects, etc.

Syndromogenesis and etiology

It is a genetic syndrome and is transmitted by autosomal recessive inheritance.

Heterozygous women frequently present: obesity, hypertension, diabetes mellitus and kidney diseases.

The basic defect is unknown.

Literature

Bardet, G.: Sur un syndrome d'obésité congenitale avec polydactylie et rétinite pigmentaire. Therese de Paris, 1920.

Biedl, A.: "Retinits pigmentosa: Ein Geschwisterpaar mit adiposogeni- taler Dystrophye." Deutsch. Med. Wschr., 48:1630, 1922.

Green, J. S. et al.: "The cardinal manifestations of Bardet-Biedl's syndrome, a form of Laurence-Moon-Biedl's syndrome." N. English J. Med., 321:1002-1009, 1989.

BASSEN-KORNZWEIG SYNDROME

Synonymy

S. of betalipoprotein deficiency. S. of beta-lipoproteinemia.

Syndromography

Clinic

It appears between 6 and 12 years of age. It is characterized by a picture of steatorrhea accompanied by acanthocytosis, retinitis pigmentosa and a progressive neurological picture: weakness of the limbs with areflexia and sensory ataxia (tabetic) that later adds a cerebellar component, Babinski sign. Kyphoscoliosis with pes cavus.

Paraclinical examinations

Blood., Pointed or spiny red blood cells (acanthocytes).

Erythrosedimentation. Diminished.

Serum low-density lipoproteins (LDL). Decrease nuidas.

Cholesterol. Decreased, 50 mg/dL. Triglyceides 10 mg/dL.

Phospholipids. Bass.

Betalipoproteins. You go down.

Jejunum biopsy. Mucosal structure normal, but absorptive villous cells are filled with fat and appear vacuolated when seen in paraffin inclusions.

Sural nerve biopsy. Decrease in the number of demyelinated nerve fibers.

Syndromogenesis and etiology

It is an extremely rare encephalopathy due to a disorder of lipid metabolism, it is inherited on an autosomal recessive basis and is characterized by the absence or deficiency of plasma beta-lipoproteins. The pathogenic mechanism is believed to be an inability of the cell membrane to synthesize beta apoproteins, which is a fundamental protein component of chylomicrons, low-density lipoproteins (LDL) and very low-density lipoproteins (VLDL). The absorbed lipids cannot be incorporated into chylomicrons or VLDL and LDL, so their passage through the membrane is prevented.

The mutational defect does not appear to be in the APOB gene, but in some other product necessary for APOB secretion.

Literature

Huang, L.S. et al. : *Exclusion of linkage between the human apolipoprotein B gene and abeliproteinemia." Am. J. Hum. Genet., 46:1141-1 148, 1990.

Stambury, J. B. et al.: The metabolic basis of inherited diseases. ed. 5 McGraow-Hill, New York, 1983.

BAILEY-CUSHING SYNDROME

Synonymy

S. of the cerebellar midline. S. archicerebellar. S. of the vermis. S. of the flocculonodular lobule. Medulloblastoma.

Syndromography

Clínic

This syndrome occurs in both sexes (more common in men) and more commonly in childhood. It is characterized by:
instability in balance, coordination disorders without apparent ataxia, morning headaches, repeated vomiting. It is accompanied by anorexia and weight loss, ophthalmoplegia (due to bilateral abductor paralysis) and papillary edema. Vertigo (related to position) and nystagmus frequently occur. In advanced stages, decerebrate seizures may appear.

Paraclinical examinations

Computerized axial tomography.

Skull radiology.

Lumbar puncture.

Electroencephalogram.

Arteriography.

Syndromogenesis and etiology

This syndrome is caused by tumors of the midline of the cerebellum and medulloblastomas.

The origin of this tumor has not been known: one theory is that it derives from fetal remains of the external granular layer of the cerebellum, another theory is that it derives from cellular remains of the lower medullary velum. The tumor frequently infiltrates the floor of the fourth ventricle and the cisterna magna, as well as around the spinal cord.

Literature

Bailey, P. and H. Cushing: "Medulloblastoma cerebelli. A common type of midcere bellar glioma of childhood." Arch. Neurol. Psychiat., 14:192-224, 1925.

Stroud, M. H.: "The otologist and the midline cerebellar syndrome." Laryngoscope, 77:1795-1805, 1967.

BELL SYNDROME

Synonymy

Acute afrigori facial paralysis.

Syndromography

Clínic

This syndrome is the most common form of facial paralysis. It is characterized by loss of voluntary motility of the muscles innervated by cranial nerve VII (facial nerve).

It occurs suddenly, when the patient stands up he observes facial asymmetry, sometimes preceded by vague discomfort around the ear.

The face is deviated towards the healthy side. This asymmetry increases as the mimicry muscles contract.

An important sign is obtained when the patient is instructed to close his eyes, which is impossible on the sick side.

The palpebral aperture remains ajar on the affected side (orbicularis oculi palsy), the movement of the eyeball is observed when trying to close the eye (Charles Bell sign) upwards and inwards (motor synergy). There is epiphora as a consequence of hypotonia of the orbicularis oculi and paralysis of the Horner muscle that cause the lacrimal punctum to move away from the region of the lacrimal lake, so the tear secretion stagnates (lagophthalmos) and spills outwards.

If the patient is asked to look upward, this is not accompanied by synergistic contraction of the frontalis muscle. on the affected side, the eyeball appears to be elevated more on the healthy side (Negro's bulbopalpebral or hyperkinetic sign).

There is a deviation of the corner of the mouth (one of the most classic symptoms) towards the healthy side, where it appears higher and is accentuated when the patient eats, speaks or laughs.

When you open your mouth, it is oval in shape (Pitres sign).

There is an inability to puff out the cheek, whistle or blow, air escaping through the corner of the mouth on the diseased side (buccinator palsy). The cutaneous muscle of the neck (platysma) does not contract. There is loss of conjunctival and corneal reflexes. Muscle tone is abolished in the paralyzed area.

According to a topographic criterion, different forms of peripheral facial paralysis can be distinguished:

1. Lesion located below the stylomastoid foramen: motor paralysis of an entire half of the face predominates.
2. Lesion that sits between the exit of the chorda tympani and the ramus of the stapedius: the paralysis is accompanied by loss of taste sensitivity (ageusia) of the anterior two-thirds of the ipsilateral tongue and decreased salivary secretion (hyposialia) of the submandibular gland.
3. Injury between the stapedius branch and the geniculate ganglion: facial paralysis, ageusia, hyposialia and painful hyperacusis (exaggerated perception of noise) due to paralysis of the stapes muscle, which produces relaxation of the tympanic membrane and consequently the vibrations caused by sound waves are large and annoying.
4. Lesion between the geniculate ganglion and the internal auditory canal: paralysis is accompanied by hyposialia, but with preservation of taste sensitivity.
5. Injury that occurs between the canal and the apparent origin of the nerve (inferior pontine sulcus): paralysis or prosoplegia may be accompanied by paralysis of other neighboring cranial nerves (trigeminal neuralgia), there is preservation of taste and decreased salivary and tear secretion.

Paraclinical examinations

Cerebrospinal fluid. Mild pleocytosis.

Electromyography. Denervation after 10 days indicates that there has been axonal degeneration.

Syndromogenesis and etiology

Its pathogenesis is unknown. It is the most frequent of paralysis, it affects adults, young and old alike.

It is proposed that in circumstances such as: traveling in vehicles with the windows open, the cold would have an important role in producing intumescence of the periosteum in the facial canal (fallopian canal) or that of the connective tissue of the nerve activated by the cold. This could also explain the seasonal or epidemic form of this disorder.

Literature

Asburg, A.K. And R.W. Guilliat: Peripheral nerve disorders. Butterworth's International Medical Review. Neurology, vol.4, Stoneham. H. A. Butterworth, 1984.

Bell, C.: "On the nerves of the face; being a second paper on that subject." Med. Classics, 1:155-169, 1936.

Groves, J.: "Bell's facial palsy in Scientific Foundations of Otolaryngology." R. Hinchcliffe, D. Hamsson (eds.). Heinemann, London, 1976. pp: 446-459.

Hauser, W. A., et al.: "Incidence and prognosis of Bell's palsy in the population of Rochester, Minnesota." Mayo Clin. Proc., 46: 258, 1971.

Karnes, W. E.: Diseases of the seventh cranial nerve. In Peripheral neuropathy, ed. 2, P.J. Dick et al. (eds). Saunders, Piladelphia, 1984, pp: 1266-1299.

Shaumberg, H.H.: P.S. Spencer and P.K. Thomas: Disorders of peripheral nerves. Davis, Philadelphia, 1983.

BENEDIKT SYNDROME

Synonymy

S. de Benedikt lower. S. of the mesencephalic tegmentum. S. posterior peduncular or peduncular cap.

Syndromography

Clínic

It is characterized by a complete hemiplegia on the opposite side of the lesion (due to seizure of the fibers of the pyramidal bundle) that is accompanied by hemiataxia, intentional or parkinsonian hemitetremor (characteristic element); hemiasynergy and other cerebellar signs. Hemianesthesia exists on the hemiplegic side.
There is paralysis of cranial nerve II (ocular motor) that is manifested by miosis, eyelid ptosis, strabismus. Hemiplegia may or may not exist when the lesion is limited to the peduncular cap.

Syndromogenesis and etiology

It is caused by a lesion of the mesencephalic tegmentum that includes the lower portion of the red nucleus, the fibers of the pyramidal bundle that have not yet crossed, the cerebellar fibers and the radicular fibers of the cranial nerve located inside the pyramidal bundle at the foot of the peduncle.

The causes that can produce it are vascular, as a complication of high blood pressure and/or atherosclerosis;
Tumors and other conditions can also cause it.

Literature

Benedikt, M.: "Tremblemen avec paralysie croisée du moteur oculaire commun." Bull. Med. Paris, 3:547-548, 1889.

BERNHARDT-ROTH SYNDROME

Synonymy

S. of meralgia paresthetica. S. of cutaneous neuritis. S. of lateral paresthesia.

Syndromography

Clinic

In this syndrome there is sensory mononeuritis that mainly affects men, its highest incidence is in doctors and is characterized by paresthetic discomfort that causes a burning sensation, stinging, tingling and slight sensitivity of the skin to the friction of clothing, the most characteristic being that it appears when the patient gets up from a chair.

Sometimes there is a burning pain of considerable intensity.

Physical examination. A region of hyperesthesia or hypoesthesia to touch, pain, and temperature is found in the corresponding external cutaneous femoral region.

Syndromogenesis and etiology

It occurs due to compression or irritation of the lateral femoralcutaneous nerve due to toxic, obese, ity, diabetes and post-surgical processes. It can also be secondary to retroperitoneal tumors.

Literature

Bernhardt, M.: "Ueber isoliert in Gebiete des N. cutaneus femoris externus vorkommende Parasthesien." Neurol. Centralbl., 14:242-244, 1895.

Dick, P.J. et al. (eds.): Peripheral Neuropathy. ed. 2, Saunders, Philadelphia, 1984.

Ecker, A.D. and H.W. Woltman: "Meralgia paresthetica. A report of one hundred and fifty cases." JAMA, 110, 1650, 1938.

BING-NEEL SYNDROME

Synonymy

S. de Waldenström. S. of macroglobulinemia.

Syndromography

Clínica

Bing-Neel syndrome constitutes a neuropathy associated with paraproteinemia. It is a complication of Waldenström macroglobulinemia.

To the symptomatology of the underlying disease, there is added a diffuse slowing of cerebral and retinal circulation that is manifested by episodes of confusion, coma and sometimes stroke, as well as peripheral neuropathy with involvement of multiple nerve trunks in most cases limited to the feet and legs with damage to tactile and vibratory sensitivity (palesthesia) and position sense (bathyesthesia), with mild ataxia and hyporeflexia.

Paraclinical examinations

Blood. The characteristic tests of macroglobulinemia are added to the findings of the neurological lesion.

Cerebrospinal fluid. Elevated proteins with increased globulins.

Syndromogenesis and etiology

Observations of familial aggregation suggest the existence of genetic and immunological factors, which in turn are related to the development of lung adenocarcinoma, lymphocytic or histiocytic lymphomas, and clinical or subclinical autoimmune diseases.

The basic defect is unknown.

Literature

Bing, J. and A.V. Neel: "Two cases of hyperglobulinemia with affection of central nervous system on a toOxi-infectious basis.
Acta Med. Scand., 88:492-506, 1936.

Bing, J.; M. Fog. and A.V. Neel: “Reports of a third case of hyperglobulinemia with affection of central nervous system on toxi-infectious basis." Acta Med. Scand., (41:409-427, 1937.

Blattner, W. A. et al.: "Waldenström's macroglobulinemia and autoimmune disease in a family." Ann. Intern. Med., 93:830- -832. 1980.

BOGORAD SYNDROME

Synonymy

S. of crocodile tears. S. of paroxysmal crying.

Syndromography

Clinic

In this syndrome, there is unilateral crying when chewing or putting a strong-tasting food in the mouth. It is characterized by the fact that mechanical stimulation and chewing without food do not produce crying.

Syndromogenesis and etiology

It is a sequel to facial paralysis and is due to an aberrant regeneration of the facial nerve fibers. In these cases the parasympathetic salivatory fibers accompany the facial nerve.

Literature

Bogorad, F. A.: "Sympton of crocodile tears." Vrach. Delo, 11:1328-1330, 1928.

BONNIER SYNDROME

Synonymy

S. of Deiter's nucleus. S. of the lateral vestibular nucleus.

Syndromography

Clínica

In this syndrome, central paroxysmal vertigo with balance disorders occurs, associated with ocular paralysis and trigeminal neuralgia.

Paraclinical examinations

Audiogram.

Angiography.

Electroencephalogram.

Syndromogenesis and etiology

Bonnier syndrome is due to vascular neoplasms with damage to the lateral vestibular nucleus (nucleus of Deiter) or the vestibular tract of the cranial nerve (vestibulocochlear).

Literature

Bonnier, P.: "Syndrome du noyau de Deiters." Compt. Yield. Soc. Biol., Paris, 4:1525-1 528, 1902.

BORRIES SYNDROME

Synonymy

S. de Symond. S. de Quinke. Symond otitic hydrocephalus.

Syndromography

Clinic

It occurs in both sexes, more common in children. It originates as a complication of otitis, due to thrombosis of the lateral sinus and is characterized by a syndrome of intracranial hypertension caused by difficulty in the resorption of cerebrospinal fluid, which causes: headaches, dizziness, vomiting, mental stupor, papillary stasis that may be accompanied by neurological signs such as conjugate deviation of the head and eyes, aphasia and homonymous lateral hemianopsia.

Paraclinical examinations

Skull radiology. Mastoid trabecular pattern disorders; sometimes widening of the cranial sutures.

Perforated tympanic membrane with purulent discharge (more common on the right side).

Blood. Leukocytosis and accelerated erythrocyte sedimentation.

Lumbar puncture. Increased pressure of the cerebrospinal fluid.

Syndromogenesis and etiology

This syndrome is due to middle ear infection and mastoiditis with lateral sinus obstruction. It occurs more frequently on the right side.

Literature

Borries, G.VT.: "Otogene encephalitis Soc. danoise laryngology." doto- Feb. 2, 1921; and Zschz. Ges. Neur. Psychiat., 70:93-101, 1921.

BOURNEVILLE SYNDROME

Synonymy

Sherlock epiloia. Tuberous sclerosis. Brushfield-Wyatt disease. Virchow scleroma. Pringle sebaceous adenomas.

Syndromography

Clínica

It is characterized by a pathognomonic symptomatic triad: imbecility, epilepsy and facial sebaceous adenomas. In addition, there is mental retardation accompanied by convulsive seizures and myoclonic seizures, with infantile spasms. Nodules the size of a pinhead and yellow in color, located in the paranasal, nasolabial and cheek areas, which together look like butterfly wings. A more common skin lesion, but more difficult to identify, are hypomelanotic macules, generally oval in shape, which can be evident with the Wood's lamp and which are present in 90% of patients and appear from birth. Periungual or subungual fibromas, gingival fibromas, and angiomyolipomas can also be observed. They may coexist: cortical cysts, bone cystic lesions in the phalanges, cardiac rhabdomyomas and more rarely pulmonary cystic lesions associated with spontaneous pneumothorax.

Sometimes there are signs of intracranial hypertension. In the fundus of the eye, pigmentary alterations, angiectasias, mulberry-shaped papilla (neurocytophacoma) or astrocytic hamartomas appear in the retina.

Paraclinical examinations

Skull radiology. Multiple intracranial calcifications; osteoporosis.

Computerized axial tomography. Calcified nodules especially in the temporal lobes and areas adjacent to the ventricles; If they are large, they can obstruct the foramen of Monro and cause unilateral or bilateral hydrocephalus. Hypodense areas may appear at the cortical junction with the white matter.

Electroencephalogram. Abnormal but without a pattern specific.

Cerebrospinal fluid. In rare cases the total protein concentration is elevated.

Syndromogenesis and etiology

Bourneville syndrome is a hamartomatous disorder that is transmitted by autosomal dominant inheritance in one third of families. The gene is assumed to be located at 9q32-q34, but the association of ET and translocation t(3;12)(p26.3; q23.3) suggests the possibility that the gene is located at 3p26 or 12q23; This supposes genetic heterogeneity. There is evidence of large variation in gene expression and reduced penetrance.

The mutation rate is 2.5/100,000 gametes. Affected siblings and healthy parents, and no family history, suggest the possibility of germline mosaicism.

Literature

Adams, R. D.: Neurocutaneous diseases in Dermatology in General Medicine. ed. 3, T.B. Fitzpatrick et al. (eds.). McGraw-Hill, New York, 1986.

Bourneville, D. M. and E. Brissaud: "Encephalite ou sclerose tubereuse des circonvolutions cérébrales." Arch. Neurol. (Paris), 1:397-4 12, 1880-1881.

Cassidy, S. B., et al.: "Family studies in tuberous sclerosis: evaluation of apparently unaffected parents." J.A.M.A., 249:1302, 1983

Ebb, D.W. and J. P. Osborne: “Non-penetrance in tuberous sclerosis." J. Med. Genet., 28:417-419, 1991.

Fahsold, R. et al. : "Tuberous sclerosis in a child with de novo translocation t(3; 12)(p26.3;q23.3)". Clin. Genet., 40:326-328, 1991.

Fitzpatrick, T.B.; G. Szabo; Y. Hori et al.: "White leafshaped macules, earliest visible sign of tuberous sclerosis." Arch. Dermatol., 98:1, 1968.

Gómez, M. R. (ed.): Tuberous sclerosis. Raven Press, New York, 1979.

Haines, J. L. et al.: "Localization of oncogene for tuberous sclerosis within 9q32-9q34 and further evidence for heterogeneity." Am. J. Hum. Genet., 49:764-772, 1991.

Janssen, W. et al.: "Genetic heterogeneity in tuberous sclerosis." Genomics, 8:237-242, 1990.

PARESTHETIC STATIC BRACHIALGIA SYNDROME

Synonymy

S. of Wartenberg. Sensory neuritis. S. with tired arms.

Nocturnal paresthetic brachialgia.

Syndromography

Clínica

This syndrome predominates in women during the climacteric and mainly affects the sensory nerves.

It is characterized by nocturnal pain in one or both arms, preferably in the dominant arm, preceded by acroparesthesia and local heat, followed by aggravating pain that does not always subside when standing up and making movements or using massages.

Paraclinical examinations

Radiology of the cervical spine and shoulders.

Electromyography.

Syndromogenesis and etiology

The idiopathic form of this syndrome is rare. The most common causes are compression or irritation of the brachial plexus.

Literature

Wartenberg, R.: "Brachialgia statica paresthetica (nocturnal arm dysesthesias)." J. Nerv. Ment. Dis., 99:877-887, 1944.

BROWN-SEQUARD SYNDROME

Synonymy

S. of hemiparaplegia. S. of spinal hemiplegia.

Syndromography

Clínic

True hemisection of the cord is exceptionally observed clinically. It is only seen in all its purity in experiments carried out on animals, since it is difficult for any of the usual causes of the syndrome to be located exactly in one half of the spinal cord.

The syndrome is manifested by motor paralysis and causes: Ipsilateral hemiplegia or monoplegia (lower limb) due to injury to the pyramidal tract, with signs of pyramidalism: deep hyperreflexia, spasticity, clonus and Babinski. Below the sectioned point there is deep anesthesia: bathyesthesia and palesthesia (mainly) ipsilateral due to seizure of the posterior cord (bundle gracillis and cuneatus) accompanied by epicritic tactile hypoesthesia, given that the fibers of this sensitivity run through the posterior cord.

There is loss of sweat secretion and the anserine reflex in the region paralyzed by injury to the vegetative pathways. At the upper limit of the lesion there is a transverse strip of total anesthesia, which surrounds half of the body due to damage to the posterior roots on the side of the injury and above this there is sometimes a small strip of hyperesthesia (due to irritation of the supra-jacent roots).

There is also loss of contralateral thermal and pain sensitivity due to injury to the lateral spinothalamic tract, coming from the opposite side of the injury; Sometimes protopathic tactile sensitivity may be affected due to damage to the anterior spinothalamic tract.

Paraclinical examinations

Spine radiology. It may show lytic, blastic changes or fractures.

Computerized axial tomography, nuclear magnetic resonance and myelography. They continue to be the fundamental examinations to demonstrate compression of the

Syndromogenesis and etiology

This syndrome is due to spinal compression due to: vertebral fractures, inflammatory, degenerative or tumor processes. It can also be caused by stab injury, syringomyelia, plaque sclerosis, etc.

The lesion affects only one half of the spinal cord, affecting the roots of the corresponding half of the segment, the pyramidal bundle, the posterior cord, the fibers of the direct spinocerebellar bundle, coming from the nuclei (Clarke's columns) located at the base of the posterior horn, and the fibers of the lateral spinothalamic bundle.

Literature

Brown Séquard, Ch. E.: Dela transmission par la moelle épiniere." C. Rend. Soc. Biol., 2:33-34, 1850.

Greenberg, A. et al.: "Epidural cord compression from metastatic tumor results with a new treatment protocol.," Ann. Neurol.. 8:361, 1980.
Tower, S. S.: “Pyramidal lesion in the monkey." Brain, 63:36, 1940.

BRUNS SYNDROME

Synonymy

S. of postural changes.

Syndromography

Clínica

In this syndrome, attacks of vertigo, headaches and vomiting appear suddenly with postural changes of the head (extension is more likely than flexion). There are no symptoms during the periods between attacks. There is irregular breathing and occasionally syncope with apnea. There is also constant head flexion or lateral flexion. The muscles in the neck region are firmly contracted and tachycardia is common.

Paraclinical examinations

Skull radiology.
Cerebral angiography.
Computerized axial tomography.

Syndromogenesis and etiology

Bruns syndrome is due to an organic lesion of the fourth ventricle or adjacent structures, caused by:
tumors, obstruction of the flow of cerebrospinal fluid or disorders of vestibular mechanisms.

Literature

Bruns, O.: “Neuropathologische Demonstrationen.” Neurol. Centrabl., 21:561-567, 1902.

CANAVAN SYNDROME

Synonymy

S. de Van Bogaert-Bertrand. Spongy degeneration of the white matter.

Syndromography

Clinic

Canavan syndrome appears in childhood (in the first three months of life). It is a condition characterized by loss of vision and optic atrophy accompanied by absence or rapid regression of psychomotor development: lethargy, sucking disorders, irritability, decreased motor activity, spastic paralysis of the limbs, increase in the size of the head (mnegaloencephaly) with separation of the sutures of the skull; sometimes seizures.

Paraclinical examinations

Cerebrospinal fluid. Normal or slightly elevated proteins.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance mainly among Jewish children. Spongy degeneration of the deep layers of the cerebral cortex and subcortical white matter, absence of myelin and hyperplasia of Alzheimer's type II astrocytes.

The basic defect appears to be related to deficiency of the enzyme aspartoacylase and high concentrations of N-acetylaspartic acid in the cerebrospinal fluid, urine, and plasma of affected individuals.

Literature

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Canavan, M.M.: "Schilder's encephalitis periaxialis diffusa, report of a case in a child aged sixteen and one-half months." Arch. Neurol. Psychiat., 25:299-308, 1931.
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NEOPLASTIC MENINGEAL CARCINOMATOSIS SYNDROME

Synonymy

S. of metastatic meningeal hypoglycorrhachia.

Syndromography

Clínica

It is characterized by polyradiculoneuritis associated with a meningeal syndrome. It usually starts in the lower limbs and is established progressively. There are sensory disorders: paresthesias, radiculalgias (frequently sciatica) accompanied by hypoesthesias on some occasions. There are motor symptoms manifested by flaccid paraplegia, areflexia and amyotrophy. Sphincter disorders frequently exist.
There is also intake of the cranial nerves: optic, oculomotor, trigeminal, facial and vagus.
Ocular paralysis (Garcín syndrome) is more common.

Meningeal syndrome does not present in its complete form, it is generally afebrile (fever only appears in late periods).

Paraclinical examinations

Cerebrospinal fluid. Lymphocytosis and increased albumin. Hypoglycorrhachia.
Neoplastic cells.

Syndromogenesis and etiology

This syndrome is due to metastasis of a primitive tumor of the breast, stomach, bronchi, colon, etc.

CARNOSINEMIA SYNDROME

Synonymy

S. of carnosinase deficiency. S. of hyperbeta carnosinemia.

Syndromography

Clínica

This syndrome occurs in the first month of life and is characterized by myoclonic seizures and psychomotor retardation.
severe.

Paraclinical examinations

Cerebrospinal fluid. High concentrations of homocarnosine.

Blood. There is elevated carnosinemia in the plasma.
Urine. Carnosine excretion.

Syndromogenesis and etiology

Carnosinemia syndrome is inherited in an autosomal recessive manner. Apparently it is an innate disorder of amino acid metabolism and is caused by a deficiency of carnosinase (aminoacylhistidine dipeptidase).

Carnosine is an alanylhistidine base contained in normal muscle tissue.

Homocarnosine is a dipeptide (gamma-aminobutyrylhistidine) found in nerve cells.

Both bases are degraded by carnosinase, which is deficient in this syndrome, which causes its increase either in the blood and urine or in the cerebrospinal fluid.

Literature

Perry, T. L.: S. Hansen: B. Tischler et al.: "Carnosinemia: a new metabolic disorder associated with neurological disease and mental defect." N. Engl. J. Med., 277:1219-1227, 1967.
Stambury, J. B. et al (eds.): The metabolic basis of inherited diseases. ed. 5 McGraw-Hill, New York, 1983.

CEREBELLAR CATALEPSY SYNDROME

Syndromography

Clinic

This syndrome was described by Babinski and is observed along with the typical symptoms and signs of cerebellar lesions.

When the patient, lying in bed in a supine position, has his thigh flexed at a right angle over the pelvis and leg, after a certain time he presents fatigue, which is manifested by oscillations and jerks and later the leg remains motionless in flexion and is able to hold it like this for a longer time than the healthy individual, who after small jerks lowers the leg again to the horizontal plane.

Syndromogenesis and etiology

Cerebellar catalepsy is due to a lesion in the cerebellum.

Literature

Babinski, J.: “De léquilibre volitionnel statique et de léquilibre volitionnel cinetique." Rev. Neurol., 10:470-474, 1902.

CAUSALGIA SYNDROME

Syndromography

Clínic

Causalgia is a rare type of peripheral neuralgia that is characterized by spontaneous, intense and diffuse pain with a burning sensation in the palm of the hands or soles of the feet, with great sensitivity of these regions to contact with clothing or air currents, heat or cold or intense noises that can exacerbate it. The patient keeps the affected limb protected and still, often wrapped in clothing moistened with cold water.

It is almost always accompanied by trophic and vasomotor disorders of the integuments, thin and smooth skin (glossy-skin), moist, with exaggerated growth of hair and nails.

Syndromogenesis and etiology

Causalgia is due to partial injury to a peripheral nerve (median or ulnar and sometimes sciatic). It is very common in times of war.

It appears that pain is produced through a short circuit of efferent sympathetic impulses to the somatic sensory fibers at the point of nerve injury, which would explain not only the vasomotor and sweating anomalies, but also the exacerbations of pain with any form of emotional stimulus. It is possible that the following three factors are present in its pathogenesis:

1. Irritive action of the post-traumatic scar on the nerve fibers included in it.
2. Irritant sympathetic injury.
3. Psychopathic predisposition of the patient.

Literature

Mitchell, S.W.: Injuries of nerves and their consequences. Lippincott, Philadelphia, 1872.

HORTON'S HEADACHE SYNDROME

Synonymy

Paroxysmal nocturnal headache. S. of histaminergic headache. S. of erythroprosopalgia. S. of migraine neuralgia. S. cluster headache.

Syndromography

Clínic

Horton's headache is more common in men (5:1) compared to women. It occurs after the age of 30 (80%).
It begins and ends abruptly (15 min-1 h). It increases when standing up and leaning forward.

There is intense burning pain in the frontoccipital region that is characterized by a constant unilateral orbital location (oculo-temporal or oculofrontal) and by its onset 2 or 3 hours after the patient has fallen asleep. During the sleep phase there are rapid eye movements. It's weird in waking hours.

During the crisis the affected side appears red, hot and sweaty. It is not accompanied by scotomas or vomiting or nausea, but it is accompanied by rhinorrhea and nasal obstruction. There is no family history. It occurs with seizures (daily) during the night with periods of remission that can last months or years. Digestive phenomena are rare.

Attacks can be triggered by alcohol, nitroglycerin or foods containing tyramine.

Physical examination. On the surface of the skull you can see the hard arterial cord.

Syndromogenesis and etiology

Although its mechanism is still unknown, it is thought that the pain attack is due to an alteration of the unmyelinated trigeminal fibers of the trigemino-vascular system.

The painful stimulus is conducted to the nucleus of the spinal tract of the trigeminal nerve and referred as visceral pain to the somaic territory, which is innervated by exteroceptive fibers of the pair.

Subsequently, the trigeminal pathway ascends to the ventral posteromedial nucleus of the thalamus and terminates in the cerebral cortex.

bral. These trigeminal fibers establish connections in the brain stem with the nucleus and parasympaic pathways of the facial nerve (greater petrosal nerve and pteryphopalatine ganglion), which would explain the parasympathetic symptoms during the headache attack (miosis, unilateral rhinorrhea, tearing, redness and sweating of the face).

On the other hand, it is proposed that the stimulation of the trigeminal fibers causes a release of neuropeptides mediated by serotinergic receptors, which increases the level of the peptide related to the calcitonin gene in the external jugular vein, producing inflammation of the serosa of the vessel wall and the dura mater and vasodilation. Inflammation of the wall of the internal carotid artery inside the cavernous sinus makes its drainage difficult, causing engorgement, or which would explain some of the symptoms and signs (stabbing pain behind the eye and in 20% of cases, Horner's syndrome). It has been suggested that the dilated internal carotid artery compresses the pericarotid fibers against the rigid bony canal, causing the sympathetic signs that accompany the attack of this type of headache.

The launch of all these mechanisms occurs periodically, suggesting that this may be due to damage to nerve centers related to vital rhythms.

Literature

Vallery-Radot, P. and P. Blamoutier: "Syndrome de vasodilatation hemicephalique d'origine sympathique (hemicranie, hemihydrorrhea, hemilarmoiement)." Bull. Mem. Soc. Méd. Hop. Paris, 125, pp. 1488-1493.

CESTAN-CHERNAIS SYNDROME

Synonymy

S. of Cestan II.

Syndromography

Clínic

This syndrome is an incomplete form of Babinski-Nageotte syndrome, a combination of Avellis and Babinski-Nageotte syndrome.

It is characterized by unilateral direct velopalatine and vocal cord paralysis on the same side (lesion of the nucleus ambiguus), contralateral pyramidal hemiplegia and hemianesthesia (lesion of the pyramidal bundle and Reil's band) of syringomyelic type. Direct cerebellar hemi-syndrome: hemiasynergy to classical tests, lateropulsion (injury to the inferior cerebellar peduncles).

It may be associated with Claude Bernard-Horner syndrome: enophthalmos, eyelid ptosis, nystagmus and ipsilateral miosis (due to injury to the sympathetic fibers of the reticular formation).

Syndromogenesis and etiology

Cestan-Chernais syndrome is caused by an injury to the medulla oblongata that affects the pyramidal tract, the ribbon of Reil, the nucleus ambiguus (somaticomotor nerves IX, X and XI), the inferior cerebellar peduncles and the substantia reticula.

The most frequent causes are: ascending infectious or degenerative processes, vascular and tumor lesions.

Literature

Cestan, R. J. and J. Chernais: “Du myosis dans certaines lésions bulbaires en foyer (hémiplégie du type Avellis associée au syndrome oculaire sympathique).” Gaz. d. hop., 76:1229-1233, 1903.

CHARCOT-MARIE-TOOTH SYNDROME

Synonymy

S. of peroneal muscular atrophy. S. of progressive peroneal muscular atrophy. S. of progressive muscular neural atrophy. S. of Tooth.

Syndromography

Clínic

Symptoms begin at puberty or early adulthood. There is weakness and paresthesia in the legs, late weakness and atrophy of the hands, slow and progressive atrophy of the peroneal muscles and other leg muscles (stork legs), progressive atrophy of the hands and arms.

Good development of shoulders, hips and trunk. Loss of deep sensitivity and reflexes in the affected parts.

Paraclinical examinations

Electromyography and nerve conduction. They show alterations.

Cerebrovascular fluid. Normal, sometimes increased proteins.

Syndromogenesis and etiology

This syndrome is characterized by genetic heterogeneity. Autosomal dominant (3.6 per 100,000) and recessive (1.4 per 100,000) inheritance has been reported, as well as dominant and recessive X-linked inheritance (3.6 per 100,000).

Although the basic defect is not well known, it appears to be the result of primary axonal degeneration affecting the roots of spinal nerves, especially motor nerves in the distal part of the limbs.

There are several locations on the X chromosome (Xp22.2, Xq26).

The gene for the dominant form has been mapped to Xp13, which shows the great genetic heterogeneity involved.

There is demyelination of the peripheral nerves (tibial and peroneal) and degenerative changes of the cells of the anterior horns and dorsal columns.

Literature

Beckett, J. et al.: "A linkage study using DNA markers localizes the gene for X-linked dominant Charcot-Marie-Tooth neuropathy." Am. J. Hum. Genet., 48: 1075-1083, 1991.

Charcot, J. M. and Marie, P.: "Sur une forme particuliere dárophie musculaire progressive, souvent familiare debutant pas lespieds et les jambes et atteignant plus tard les mains." Rev. Med., 6:97-138, 1886.

Dick, P. J. and E. H. Lambert: "Lower motor and primary sensory neuron diseases with peroneal muscular atrophy." Arch. Neurol., 18:603-618, 1968.

Tooth, H. H.: The peroneal type of progressive muscular atrophy. Lewis, London, 1886.

CHARCOT-WEISS-BAKER SYNDROME

Synonymy

S. of the carotid sinus. S. of vagal syncope. S. de Weiss-Baker. S. vaso-vagal. S. de Gowers. DaCosta S. S. of cardiac neurosis. S. of neurocirculatory asthenia. S. vasovagal.

Syndromography

Clinic

It is the most common form of syncope, it occurs at any age, with precipitating factors that include: fatigue, emotion, some trauma with pain, lack of food or sleep, indigestion, closed environment, seeing blood, or many times no apparent factor.

The patient, before syncope, is standing or sitting, never lying down. Premonitory symptoms: sudden weakness, sweating, vertigo, epigastric discomfort, paresthesias, palpitations, sialorrhea.

Syncope can be prevented by putting the patient to bed.

Syncope is manifested by loss of consciousness for a few seconds or minutes, occasionally accompanied by tonic-clonic movements; there is recovery without sequelae, nervousness, vertigo, headache may occur.

Some patients may suffer all symptoms without complete loss of consciousness.

This syndrome occurs more frequently in males. It has a sudden onset and is characterized by generalized muscle weakness, loss of postural tone, inability to remain erect, and loss of consciousness.

Two types can be recognized:

1. Vagal type. It is characterized by sinus bradycardia, sinus arrest and even atrioventricular block.
2. Depressant type. Decrease in blood pressure without decrease in heart rate.

Both types can coexist.

Physical examination. Paleness, coldness, sweating, moderate cyanosis, weak pulse. Bradycardia (sometimes preceded by tachycardia), hypotension.

Paraclinical examinations

Electroencephalogram. It shows high-voltage, slow waves of 2-5 s that coincide with loss of consciousness.

Special tests. Rule out epilepsy or heart disease.

Syndromogenesis and etiology

Charcot-Weiss-Baker syndrome is caused by sudden movements of the head to the sides, by wearing a tight neck, or by shaving the carotid sinus region. The lack of such stimuli does not rule out the diagnosis, as it can occur spontaneously.

It can also be caused by atherosclerosis. Other forms of vasovagal syncope have been described secondary to intense pain of visceral origin, injury to the esophagus or mediastinum, pleural or peritoneal puncture, and intense dizziness.

The carotid sinus is normally sensitive to stretch and gives rise to sensory impulses that are conducted by Hering's nerve (branch of the glossopharyngeal nerve) to the medulla oblongata. It is important to distinguish between a benign disorder (carotid sinus hypersensitivity) and more serious disorders, such as atheromatous narrowing of the carotid sinus.

The loss of consciousness is due to less oxygenation of the areas of the brain on which consciousness depends, caused by a significant decrease in cerebral blood flow, the use of oxygen by the brain and vascular resistance at that level. The sudden, reflex loss of peripheral resistance temporarily creates cerebral anoxia. It can also be triggered by a reaction to cold.

It is especially observed in young people with an asthenic habit and neurovegetative imbalance.

Literature

Charcot, J. M.: Lecons sur les maladies du systeme nerveux faites a la salpetriere. Paris, 1872-1873.

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Foster, M.: Text book of physiology. MacMillan, London, 1888, Pp. 297;345.

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Wayne, H. H.: "Syncope: physiological considerations and an analysis of the clinical characteristics in 510 patients." Amer. J. Med., 30:418-438, 1961.

Weiss, S. and J. P. Baker: The carotid sinus reflex in health and disease. Its role in the causation of fainting and convulsions." Medicine, 12:297-354, 1933.

CHARCOT-WILBRAND SYNDROME

Syndromography

Clinic

Visual agnosia and agraphia sometimes associated with Gertsmann syndrome occur.

Syndromogenesis and etiology

This syndrome is caused by injury to the angular gyrus artery on the dominant side.

Literature

Charcot, J. M.: Sur un cas de cecité verbales. Ouvres completes de Charcot. Delahaye Lecrosnier, Paris, 1887.

CLAUDE BERNARD-HORNER SYNDROME

Synonymy

S. of paralysis of the cervical sympathetic system. S. of the stellate ganglion. S. oculopupillary. S. de Bernard-Horner. Horner's S.

Syndromography

Clinic

This syndrome presents a clinical picture characterized by miosis, enophthalmia, blepharoptosis, hypotonia of the eyeball with congestion of the conjunctiva and narrowing of the palpebral fissure.

Paradoxically, mydriasis may occur after a few days, due to a great psychological disorder or stress. Eyelid ptosis, apparent or with minimal exophthalmos, ocular hypotonia.
Increased temperature on the ipsilateral side of the face. Increase or decrease in tearing. Decreased sweating of the face (injury of the common carotid artery below the bifurcation). Occasionally cataract development. In children there may be depigmentation of the iris.

Paraclinical examinations

Failure of pupillary dilation to the action of this drug.
Test with adrenergic substances. Exaggerated response.
Brain and thoracic computed axial tomography.
Angiography.
Chest radiology with technique to study vertex.

Syndromogenesis and etiology

Claude Bernard syndrome is caused by apical pleuropulmonary lesions, whether tumorous, infectious, or supernumerary cervical ribs.

It is due to a paralysis of the upper cervical sympathetic nerve due to injury to the oculo-pupillary sympathetic fibers in any part of the path, from its ciliospinal center of Budge (TI) to the long ciliary nerves passing through the white rami communicans (CVIII-TI), stellate ganglion, middle and superior cervical ganglia.

It can be caused by: trauma, surgery, neoplasms, thrombosis or aneurysms.

Literature

Bernard, C.: "Recherches expérimentales sur le grand symptahique et especiallyment sur línfluence que le section de ce nerf exerce sur la chaleur animal." Compt. Yield. Soc. Biol. (Paris), pt. 2, 5:77, 1853.

Horner, E: "Ueber eine Form von Ptosis." Klin. Mbl. Augenheilk., 7:193-198, 1862.

Langham, M.E. and G.W. Weinsteim: "Horner's syndrome. Ocular supersensitivity to adrenergic amines." Arch. Ophthal., 78:462-469, 1967.

COGAN II SYNDROME

Synonymy

Cogan's oculomotor apraxia. S. of congenital absence of lateral gaze.

Syndromography

Clinic

It is observed only in children. The patient is unable to turn his eyes to the side on command. When turning to one side, the child turns his head towards the desired side but his eyes are delayed and turn to the left, therefore, he has to surpass the desired target with his head, in order to be able to fix his gaze forward. Once the gaze has been fixed on the focused object, the head returns to its original position. In a compensatory manner, the patient develops head movements that characterize his attempts at voluntary looking. These patients often take longer to learn to walk.

Paraclinical examinations

Caloric stimulation of the labyrinth. It causes tonic movements (cold towards the side of the stimulus, hot towards the opposite side), but not nystagmus as in a healthy person. It is impossible to induce optokinetic nystagmus.

Syndromogenesis and etiology

It is a syndrome of genetic etiology and autosomal recessive inheritance. The basic defect is unknown.

Literature

Cogan, D. C.: "A type of congenital ocular motor apraxia presenting jerky head movements." Tr. Am. Ophthalmol., 56:853, 1952.

HORSE TAIL SYNDROME

Synonymy

S. of the tumor of the filum terminalis. S. of the cauda equina tumor. S. of intermittent claudication of the cauda equina. S. of pseudoclaudication.

Syndromography

Clínica

The most characteristic of this syndrome are headaches.

root type. Complete peripheral paralysis of the lower limbs (involvement of the last four lumbar roots and all the sacrococcygeal roots). The anesthesia area can affect the entire lower limb without reaching the inguinal fold and testicular sensitivity is respected (the first lumbar root is not taken).

This association of anesthesia and pain (painful anesthesia) is characteristic of these injuries, unlike injuries limited to the conus medullaris, which are not accompanied by pain.
Sphincter disorders, degenerative atrophy and fibrillation, hypotonia and abolition of patellar and Achilles reflexes, and genital impotence are present.

It is the only peripheral paralysis that is accompanied by sphincter disorders. There is no Babinski sign.

Paraclinical examinations

Lumbar puncture. Early blockage of the cerebrospinal fluid (positive Queckenstedt test) and high protein content.

teico lean towards an extramedullary location.

Contrast myelography. It constitutes the most important diagnostic method and, together with the study of cells in the cerebrospinal fluid with a millipore filter, are the most useful laboratory methods.

Selective spinal angiography.

Syndromogenesis and etiology

The most common causes are: tumors, trauma or other compressive causes in the cauda equina region. Compression of the L2-L5 nerve roots and all sacrococcygeal roots.

Literature

Déjerine, J.: "La claudication intermittente de la moelle épiniere." Presse Med., 2: 981-984, 1911.

Hardy, A. G. and A. B. Rossier: Spinal cord injuries: orthopedic and neurological aspects. Stuttgart, Thieme. 1975.

Kavanaugh, G.J.; H.J. Svien; C.B. Holman et al.: "Pseudoclaudication: syndrome produced by compression of the cauda equina." J.A.M.A., 206:2477-2481, 1968.

BRAIN COMPRESSION SYNDROME

Syndromography

Clinic

It is the most frequent complication of severe brain trauma. It is characterized by the sudden or progressive aggravation of the symptoms, initial prolongation of the state of unconsciousness, establishment of a condition of drowsiness and progressive mental dullness that evolves into coma. Symptoms can occur even when there has been no initial loss of consciousness, after apparently trivial traumas. It is usually accompanied by intense headache, commonly generalized with frontal and occipital predominance or limited to the area related to the trauma. There is frequently projectile vomiting not preceded by nausea (control vomiting).

In addition, there is a slow pulse rate (40-50/min) and deep and irregular bradypnea with respiratory pauses (Cheyne-Stokes dyspnea). When the compression is small, the syndrome usually

be discrete and sometimes non-existent and is characterized by focal symptoms: irritation phenomena (Jacksonian or focal crises) or motor, sensitive or sensory deficits that depend on the affected area. When the compression is limited or affects silent areas such as the right frontal or parietotemporal lobe, the symptoms are minimal.

If the compression secondarily produces displacement of the brain, it may be accompanied by other symptoms. The most frequent is the herniation of the temporal lobe through the incisura of the tentorium cerebellum, which causes compression of the oculomotor, this is translated by unilateral mydriasis and pupillary rigidity associated with signs of brainstem compression: generalized hypertonia (in extension) and tonic cervical reflexes (decerebrate rigidity syndrome).

There is ipsilateral hemiplegia due to compression of the cerebral peduncle on the opposite side against the edge of the incisura, homonymous hemianopia due to compression of the posterior cerebral artery, and secondary damage to the cortical center of vision (occipital lobe).

Another secondary compression consists of herniation of the cerebellar tonsils giving severe bulbar symptoms.

Syndromogenesis and etiology

The most frequent causes of this syndrome are: meningeal or intracerebral hemorrhages, cerebral edema as a result of trauma, traumatic edema (most common cause). It can also be a consequence of hydrocephalus or a collapsed skull.

INTERMITTENT VERTEBRAL COMPRESSION SYNDROME

Synonymy

S. de Rochain-Bärtschi. S. compression of the vertebral artery. S. of cervical vertigo.

Syndromography

Clínic

This syndrome appears unexpectedly. There are predisposing factors such as emotional tensions and rotation or extension of the head. It is characterized by the presence of vertigo, accompanied by decreased hearing, tinnitus, headache, diplopia, balance disorders, and gastrointestinal symptoms: vomiting, nausea, explosive diarrhea. There are visual disorders, paresthesias, sensation of numbness and coldness of the ipsilateral limb.

Physical examination. Decreased or absent radial pulse, supraclavicular murmur (30% of cases), which appears with change in head position. Ataxia, dysarthria, nystagmus.

Paraclinical examinations

Vertebral Doppler.

Radiology. of spine with oblique views.

Vertebral angiography.

Syndromogenesis and etiology

The most common causes are: anomaly of the vertebral arterial system, which causes intermittent compression at the origin or in the course of the artery through the transverse foramina of the first six cervical vertebrae; spinal cord defect that intermittently compresses the vertebral arteries.

It becomes evident when exaggerated and forced movements of the head and neck are made (hyperextension, twisting, etc.).

Literature

Morley, J. B.: "Unrupted vertebro-basilar aneurysm." Med. J. Austr., 2:1024-1027, 1967.

Pratt-Thomas, H. R. and K. E. Berger: "Cerebellar and spinal injuries after chiropractic manipulation." J.A.M.A., 133:600- -603, 1947.

CONCUSSION SYNDROME

Syndromography

Clinic

Traumatic brain injuries are classified as: concussion, contusion and compression.

Brain concussion is the sudden suppression of brain functions that appears immediately after a head injury without apparent brain injuries.

Concussion is defined as a temporary abolition of consciousness due to a blow to the skull, it is always immediate and its effects can last a variable time (seconds, minutes, hours or more).

It is characterized by: immediate abolition of consciousness, suppression of reflexes, temporary arrest of breathing, bradycardia and hypotension, dilated pupils, abolished pupillary reflexes, cold skin, weak and rapid pulse, shallow and irregular breathing. Generally, vital signs return to normal in a few seconds while the patient remains unconscious. After a short period, the patient begins to agitate, opens his eyes but does not see, the corneal, pharyngeal and cutaneous reflexes reappear and the limbs react to painful stimuli. Little by little he recovers contact with the environment, answering simple questions but in a situation of retrograde amnesia, so he is subsequently unable to remember what he has said, in the end he achieves recovery.

The best index of the severity of the shock is the duration of the period of amnesia. Fixation memory is affected and the patient repeats the same question as if he managed to retain the answer or understand its meaning. The headache is usually constant and vomiting is not frequent. It is not uncommon for the patient to have difficulty concentrating his ideas, judging facts and making decisions for some time, gradually becoming fatigued when he tries.

Sometimes when you do not take care of yourself and do not get enough rest, the symptoms can reappear, constituting the so-called post-concussion syndrome.

If the shock is intense, it can cause death.

Paraclinical examinations

Cerebrospinal fluid

Blood.

Computerized axial tomography.

Literature

Ballet, Gilbert: Des absces du cerveau consecutifs a certains malformations cardiacaques. " Arch. Gén. de Méd., 145:659-667, 1880.

Chambers, W. R.: *Brain abscess associated with pulmonary arteriovenous fistula." Ann. Surg., 141:276-277, 1955.

Farre, JR: Pathological investigations. Essay I. On malformations of the human heart; illustrated by numerous cases, and preceded by some observations on the method of improving the diagnostic part of medicine. London, Longman, 1814.

CONUS MEDULARARY SYNDROME

Synonymy

S. sacroanogenital-vesical.

Syndromography

Clinic

In this syndrome there is flaccid paralysis of the gluteal muscles and anterolateral region of the leg. Atrophy and fasciculation may accompany the condition. Pain in the back and sciatic and perineal region. Anesthesia limited to the perineal region. Sphincter disorders: unconscious urination, ejaculation and erection abolished.

Physical examination. Plantar reflex abolished, Achilles reflex preserved. Vesicopudenda saddle anesthesia. Paralysis of the plantar flexors of the fingers.

Syndromogenesis and etiology

The causes that cause it are: tumor of the conus medullaris, trauma, compression of the S2-S5 spinal segments, located at the level of the DI12 to L1 vertebrae.

Literature

Roussy, G. and J. Lhermitte: Les blesseurs de la moelle et de la queue de cheval. Paris, 1918.

ACUTE KOREA SYNDROME

Synonymy

Sydenham Korea. S. of the dance of Saint Vitus. S. of chorea minoro rheumatic chorea.

Syndromography

Clinic

Acute chorea occurs in children almost always between 6 and 12 years of age, but can be observed between 15 and 18 years of age.
It affects the female sex more frequently.

In general it has an insidious beginning. The child first manifests changes in character, he becomes naughty, inattentive and unstable, he is frequently punished at school or at home for inappropriate attitudes (when eating he throws the cutlery, stains the notebooks when writing), he is described as nervous, careless or clumsy." After a few days, choreic movements appear, which are disordered, involuntary and of great amplitude in the upper limbs, small at first and later in the form of jerks, bruises of the fingers and hands that make any movement impossible. manual activity (writing, eating, dressing). The patient acts as a clown (Levi Valensi), walking becomes difficult and irregular, hesitating, the legs are crossed, he stumbles and cannot maintain his balance (doll-like gait).
These manifestations intensify with difficulty. emotions and may be limited to one side (hemicorea) or be generalized.

It is accompanied by a decrease in muscle strength, atony, asthenia, and the muscle masses are soft. No paralysis is observed, deep reflexes are generally preserved, but sometimes abolished. The response of the patellar or Achilles reflex is characteristic. By striking the quadriceps tendon, It produces extension of the leg that is maintained for a longer time than normal. When struck again several times, the leg can be held in extension for several seconds (Gordon's tonic reflex). The same phenomenon occurs with the Achilles reflex with persistent extension of the foot. Sometimes what occurs is the lowering of the leg in an oscillating manner (pendulum reflex) due to muscle hypotonia.

The choreic or Wilson hand is characteristic with accentuated flexion of the wrist, hyperextended fingers at the level of the metacarpophalangeal joints. When raising the hand, it is placed pronated and with the thumb projected forward.

There are constant psychological disorders that appear from the beginning of the disease, characterized by changes in character, attention disorders, restlessness, irritability, emotional instability, which are proportional to the severity of the illness.

Korea.

Choreic movements constitute the basis of the diagnosis.

Cardiovascular disorders reveal the signs present in rheumatic fever: cottoning of the first heart sound that gradually transforms into a systolic murmur (mitral regurgitation), tachycardia, extrasystolic arrhythmia, fever (on occasion).

Paraclinical examinations

AntistreptolysinO (ASO) test. Low or borderline titles.

Antistreptococcal antibodies (anti-DNAse or antihyaluronidase and antistreptoprozyme test. It is a hemagglutination reaction to concentrated extracellular antigens of streptococci absorbed into erythrocytes, it constitutes a very sensitive index of streptococcal infection and reaches titers greater than 200 /mL.

Syndromnogenesis and etiology

Acute chorea is a late manifestation of rheumatic fever. It usually appears after a long latency period (months) after a streptococcal infection and when all manifestations of rheumatic fever have disappeared.

It has been observed as an initial manifestation of various processes of epidemic encephalitis, encephalopathies due to rash, whooping cough, diphtheria, in idiopathic hypocalcemia, hyperthyroidism, generalized lupus erythematosus, carbon monoxide poisoning, vascular diseases, tumors, degenerative processes of the basal ganglia (all in a small percentage of patients).

Literature

Huntington, G.: "On chorea." Med. Surg. Reporter, 26:317-321, 1872.

HEREDITARY CHRONIC PROGRESSIVE CHOREA SYNDROME

Synonymy

Huntington's chorea or Huntington's disease. S.
chronic progressive degenerative chorea. S. of hereditary adult chorea. Hereditary St. Vitus disease.

Syndromography

Clínica

Athetotic movements and progressive dementia are associated. It generally occurs around mid-adult life (35-40), rarely in children. Imprecise start. The movements

involuntary choreic type in the head, trunk and limbs (respect the eyes), they are wide, unbalanced. Grimaces, grimaces, dysarthria, sucking and projection movements of the tongue, twisting of the upper limbs and trunk. Decreased muscle tone. Hypermetry. Conservation of deep reflexes. Gordon's sign by causing the knee jerk reflex. Characteristic dancing march.

Dementia accompanies the motor disorder, there are psychological manifestations, depression, erratic behavior and emotional crises that are sometimes the first symptoms that disable the patient, long before the motor or dementia disorders become serious.

Syndromogenesis and etiology

The lesion is located at the level of the basal nuclei of the telencephalon. There is replacement of the small cells of the striatum by fibrous astrocytes with preservation of the large cells (the anterior areas of the putamen and caudate are more affected), accompanied by alterations of the globus pallidumn, subthalamic nuclei, and the red nucleus with slight modifications of the substantia nigra. There is slight neuronal loss of layers 3, 5, and 6 of the cortex with gliosis.

Currently there is talk of biochemical disorders characterized by an increase in the amount of dopamine, which attempts to explain the involuntary movements induced by L-Dopa. It has been postulated that the involuntary movements that appear in this condition are produced by an increase in the response of striatal receptors to dopamine, supported by the induction of tardive dyskinesias in the chronic administration of phenothiazines.

On the other hand, the possibility of a disorder in the metabolism of neurotransmitters such as gamma-aminobutyric acid (GABA) and acetylcholine is raised, as a decrease in the enzymes glutamic acid decarboxylase and acetylcholine transferase, necessary for the production of these neurotransmitters, is found in the basal ganglia of these patients. The disruption of the homeostatic relationship of these agents may be the substrate of the choreic manifestations of this disease.

The etiology is genetic and is transmitted by autosomal dominant inheritance. The mutation has been located in 4p16.3. It has been detected that the gene presents an amplification of CAG repeats that never exceed 100. This repetition is translated into a polyglutamine chain. These chains are found in the structure of proteins that bind to DNA, and the extension of this polyglutamine segment more than normal can affect its biological role and explain syndromogenesis.

Another genetic factor that is invoked is genomic imprinting, which explains the anticipation of symptoms that depend on paternal transmission (if the gene is inherited paternally, the symptoms begin earlier, if it is inherited maternally, they appear later). A 50% risk is considered for the children of an affected parent.

Literature

Ferrante, R.J. et al.: "Selective sporing of a class of striatal neurons in Huntington' s disease." Science, 230: 561, 1985.

Hutington, G.: "On chorea." Med. Surg. Reporter, 26:317-321, 1872.

Martin, J. B.: "Huntington's disease. New approaches to an old problem." Neurology, 34:1059,1984.

Von Sattel, J. P. et al.: "Neuropathological classifications of Huntington's diseases." J. Neuropathol. Exp. Neurol., 44: 559,1985.

KOREA GRAVIDICA SYNDROME

Syndromography

Clínic

Chorea gravidarum is usually seen in young primiparas in the first half of pregnancy. It is characterized by very intense choreiform movements accompanied by some mental disorders. Abortion occurs in 60% of

cases.

Syndromogenesis and etiology

The etiology is unknown, but it is suggested that this syndrome may be related to attacks of rheumatic fever.

Literature

Bisno, A.: Acute rheumatic fever: current concepts and controversies. In: Swartz. M.N. and J, S. Remington (eds.).
Current clinical topics in infectious diseases. McGraw-Hill Book Co., New York, 1989. pp. 316-341.

Huntington, G.: "On chorea." Med. Surg. Reporter, 26:317- -321,1872.

SENILE KOREA SYNDROME

Synonymy

S. non-hereditary chronic progressive chorea.

Syndromography

Clinic

This syndrome is characterized by lower intensity of motor symptoms. It appears after 50 years. The accentuated psychological disorders and the absence of progressive aggravation are missing. The patient presents light movements, generally on only one side of the body that takes in the limbs, although they can be bilateral. Complex involuntary movements of the face, mouth and tongue that may be the only manifestation of the syndrome. It is not associated with a mental disorder or family history.

Syndromogenesis and etiology

Due to the cerebral changes secondary to vascular pathology, it has been suggested that this syndrome is a late-onset variant of Huntington's chorea, since it involves the caudate nucleus but the cortex is spared.

Literature

Huntington, G.: "On chorea." Med. Surg. Reporter, 26:317-321, 1872.

Martin, J. B.: "Huntington's disease: new approaches to an old problem." Neurology, 34:1059, 1984.

Weiner, W. J.: «Lingual-facial-buccal movements in the elderly.
Pathogenesis and relationships to senile chorea." J. Am. Geriat. Soc.,21:318, 1973.

CREUTZFELDT-JAKOB SYNDROME

Synonymy

S. of spastic pseudosclerosis. S. of corticostriospinal degeneration. S. of spasmodic pseudosclerosis. Sick

Jakob-Creutzfeldt age. Subacute spongiform encephalopathy. S. of Heidenhain. S. of subacute spongiform encephalopathy. S. of cortical blindness.

Syndromnography

Clínica

It is a rare process that appears in the middle age of life around 40 years, but it can occur in young adults. It begins insidiously.

It begins with behavioral changes, memory disorders and visual difficulty accompanied by hallucinations (sometimes), illusions and other manifestations of delirium followed by progressive loss of vision, progressive central mental deterioration associated with extrapyramidal symptoms: involuntary movements of choreic and athetotic type and myoclonus accompanied by pyramidal signs (less frequent) in the form of spastic paraparesis. There are mental disorders of a dementing type that develop rapidly, emaciation, generalized rigidity, stupor and coma.

Cerebellar ataxia and muscle atrophy with fasciculations are inconstant events.

Focal signs sometimes suggest the presence of an intracranial tumor. The condition is afebrile and death can occur in less than a year with an average of seven months. It is important from a clinical point of view to differentiate this condition from Alzheimer's disease and other forms of dementia, as well as from expansive-type intracranial lesions.

Paraclinical examinations

Cerebrospinal fluid. A slight increase in protein is common. Electroencephalogram. Changes throughout the development of the disease, which vary from diffuse and nonspecific to sharp and synchronous waves of high potential on an increasingly flat background.

Microbiological. Inoculation of animals from biopsy samples.

Syndromogenesis and etiology

The disease is caused by a filterable agent, with the capacity for self-replication, transmissible to chimpanzees, monkeys, guinea pigs and mice. The transmission mechanism is unknown, but several cases have been reported due to iatrogenesis (corneal transplantation and application of electrodes to the cerebral cortex after having used the instruments of these techniques in affected patients). In other cases, a history of neurological and ocular operations is recorded a few years before the onset of the disease.

It is considered that the filterable agent is probably a slow-acting virus and the incubation period may be 20 years.

The lesion consists of cortical degeneration, preferably in the occipital cortex or less generalized to other cortical areas.

Literature

Benenson, A.: The control of communicable diseases in man. Scientific Publication No. 507 PAHO/WHO 1987, p. 94.

Creutzfeldt, H.G.: Uber eine eigenartige herdformige Erkankung des Zentralnervensystems. In: Nissl, F. and A. Alzheimer: Histologie und Histopathologie, Jena, Fischer, Arbeit Erganzungbasband, 1921.

Heidenhain, A.: "Klinische und anatomische Untersuchungen uber eine eigenartige organische Enkrankung des Zentralnervensys- tems im Praesenium." Z. Neurol. Psychiat., 118:49, 1929.

BODY LOUIS SYNDROME

Synonymy

Ballism S. S. of hemiballism. S. of the subthalamic nucleus.

Syndromography

Clínic

This syndrome is characterized by involuntary movements of greater amplitude than choreic movements, called ballismos. They occur in one part of the body (hemibalismus) or in both parts. During movements the upper limbs are violently projected forward, but this is less pronounced in the lower limbs.

Syndromogenesis and etiology

This syndrome is attributed to contralateral injuries to Louis' body caused by: hemorrhages, tumors, trauma, etc.

Literature

Fisher, O.: "Zur Frage der anatomischen Grundlage der Athetose double und der posthemiplegischen Bewegungsstorung uberhaupt." Z. ges. Neurol. Psychiat., 7:463, 1911.

DANDY-WALKER SYNDROME

Synonymy

S. of atresia of the foramina of Luschka and Magendie. S. of Dandy-Walker deformity.

Syndromography

Clinic

Signs of intracranial hypertension are present: vomiting, hyperirritability, convulsions, elongation of the skull, congestion of the superficial veins of the skull, bulging anterior fontanel, separated sutures, papilledema, bradycardia and bradypnea.

Syndromogenesis and etiology

Dandy-Walker syndrome is due to a congenital malformation in which there is a cystic expansion of the IV ventricle due to the inability to open the foramina of Luschka and Magendie. This ventricle is transformed into a large dilated cavity on which the cerebellar vermis is located, the lateral sinus is elevated and the tentorium cerebellum is displaced.
The obstruction of the foramina prevents the free flow of cerebrospinal fluid from the ventricular system to the subarachnoid space, for its subsequent reabsorption, causing a great dilation above them that prevents the proper development of the cerebellar vermis.

Literature

Dandy, W.E.: "The diagnosis and treatment of hydrocephalus due to occlusions of the foramina of Magendie and Luschka." Surg. Gynec. Obstet., 32:112-124, 1921.

Walker, A. E.: "A case of congenital atresia of the foramina of Luschka and Magendie; surgical cure." J. Neuropath. Exp. Neurol., 3:368-373, 1944.

PYRAMIDAL DECUSATION SYNDROME

Synonymy

S. of crossed hemiplegia.

Syndromography

Clínic

Paralysis of an upper limb accompanied by paralysis of the contralateral lower limb that spares the face.
Spasticity, increased tone of the affected muscles, absence of the abdominal reflex on the side of the affected lower limb, hypereflexia, clonus of the knee, Babinski sign.

Syndromogenesis and etiology

This syndrome is caused by injury to the lower part of the medulla oblongata, at the specific distal point of the decussation of the fibers that innervate the upper limbs and proximal to the decussation of the fibers corresponding to the lower limb. The most common cause is trauma at that level.

Literature

Chavany, J. A.; J. N. Taptas and D. Haggenmuller: "Les faux syndrome alternes dórigine hemisphérique; l'hemiplegia cruciata' par bilateral cortical lesions." Presse Med., 60:1126-1128, 1952.

DEDÉJERINE-KLUMPKE SYNDROME

Synonymy

S. of neuritis of the brachial plexus.

Syndromography

Clínic

Condition characterized by pain, hyperesthesia or loss of sensitivity of the medial side of the hand, weakness and paralysis of the hand, accompanied by vision disorders (Claude Bernard-Horner syndrome): miosis, enophthalmia and eyelid ptosis.
Atrophy of the interosseous muscles, the thenar region, hypothenar region, flexor carpi ulnaris, common flexor digitorum and sensory disorders of the ulnar region of the upper limb.

Syndromogenesis and etiology

The syndrome is caused by injuries that affect the cervical cord in its lower part (eighth cervical and first thoracic roots) or the brachial plexus, and the sympathetic fibers.

The sympathetic nerve is affected by injury to an anastomotic branch that goes to the stellate ganglion or by direct injury to the latter.

It can be caused by a tumor of the apex of the lung, any malignant tumor that occupies the superior costovertebral sinus and compresses the brachial plexus. Trauma (50% of cases).

Literature

Déjerine Kumplle, A.: *Contribution á l'etudedes paralysies radiculaires du plexus brachial. Total root paralysis.
Paralysies radiculaires inférieures de la participation des filets sympatiques oculopupillaires dans ces paralysies." Rev. de Med., Paris, 5:591-616, 739-790, 1885.

DEJERINE-ROUSSY SYNDROME

Synonymy

S. thalamic. S. of thalamic anesthesia.

Syndromography

Clínic

It is characterized by more or less evident contralateral hemianesthesia of superficial sensitivity (tactile, thermal and painful); more or less marked and very pronounced deep sensitivity (proprioceptive): vibration and joint position (palesthesia and bathyesthesia), the stereognostic sense is completely abolished. These alterations affect the face, arm, trunk, leg and foot; sometimes only one member. There is slight and rapidly regressive contralateral hemiplegia, choreic or athetotic movements in the limbs with paresis, paroxysmal pain, sometimes intolerable that varies with changes in the affective tone (hyperpathy) that frequently spares the face. In addition, there is an elevation of the threshold of sensations.

Contralateral homonymous hemianopia may exist when the lesion is located in the pulvinar of the thalamus and in the lateral geniculate nucleus.

It is common for the hand to adopt a characteristic position, hyperextended fingers, athetoid tremor (thalamopallidal hand) accompanied by deformation of the nails and thinning of the skin.

Paraclinical examinations

Computerized axial tomography. Checks for injuries caused by lack of irrigation in the territory of the posterior cerebral artery.

Nuclear magnetic resonance. It can detect infarcts larger than 0.5 cm.

Angiography. It is the only safe method to confirm vascular etiology.

Syndromogenesis and etiology

The lesions cause involvement of the posterolateral ventral nucleus of the thalamus. The pyramidal (corticospinal) bundle that runs through the posterior arm of the internal capsule adjacent to the thalamus may be involved, which explains the condition of hemiparesis or hemiplegia. According to Lermitte, painful perception of normal stimuli is produced by loss of the protective function of the thalamus, as a brake or modulator of the afferent impulses that pass through it on their way to the cortex.

This syndrome is caused by vascular lesions of the posterior cerebral arteries (most common), gliomas, encephalitis and trauma.

Literature

Déjerine, J. et G Roussy: “Le syndrome thalamique." Rev. Neurol., 14:521-532, 1906.

DEVIC SYNDROME

Synonymy

S. of neuromyelitis optica. S. neuropticomyelitic. S. of diffuse myelitis with neuritis.

Syndromnography

Clinic

It is a rare, subacute, demyelinating condition that appears at any age (more common in childhood) and is characterized by ocular and spinal manifestations. Generally, ocular symptoms are the first, followed by a period of days or weeks of spinal symptoms.

Ocular symptoms are manifested by bulbar or retrobulbar optic neuritis (in the first the papilla is normal, in the second there is edema of the papilla). Amblyopia (bilateral central scotoma), spontaneous pain in the eyeballs that is accentuated with movements. Neuritis commonly affects one side and later (within hours or days) the other side. Spinal symptoms are characterized by transverse myelopathy: paraplegia, tetraplegia (often first flaccid and then spastic), sphincter disorders, and anesthesia below the lesion.

Paraclinical examinations

Cerebrospinal fluid. Increased albumins and mild lymphocytic pleocytosis.

Syndromogenesis and etiology

It can be considered a variant of multiple sclerosis. Its etiology is unknown.

It affects the optic nerves, chiasm and spinal cord due to intense demyelination, and various segments of the spinal cord, most common in the cervical region.

Literature

Devic, M. E.: "Myelite subabaigue compliquee de nérvite optic." Bull. Med. Par., 8:1033, 1894.

Greenfield, J. G. and R. M. Norman: Demyelinating diseases. In Blackwood, W.: Greenfield's Neuropathology. Baltimore, Williams & Wilkins, 1963.

ACUTE DISSEMINATED ENCEPHALOMYELITIS SYNDROME

Synonymy

S. de Redlich-Fatau. S. of acute multifocal myelinoclasis.

Syndromnography

Clínic

It occurs in both sexes and at any age, although it is more common in young individuals between 15-30 years old. With a sudden onset, this syndrome is characterized by fever accompanied by migraine-like headache, vertigo, nausea and vomiting, paresthesia or pain in the lower limbs or trunk, flaccid paralysis, dysphagia, dysphonia, ataxia, seizures, delirium and optic neuritis.

Paraclinical examinations

Blood. Leukocytosis.

Cerebrospinal fluid. Pleocytosis.

Syndromogenesis and etiology

This syndrome is related to viral infections (measles, mumps, chickenpox, rubella, influenza, infectious mononucleosis, smallpox and poorly characterized respiratory infections). After immunizations against smallpox, rabies, influenza or tetanus.

Its pathogenesis can be considered as a direct consequence of the viral infection, with immune disorders or as a result of the interaction of the two mechanisms.

Literature

Marburg, 0.: "Die sogenannte akute multiple Sklerose (Encephalomyelitis periaxialis scleroticans)." Jahrb. F. Psychiat., 27:213-312, 1906.

Miller, H. G. and M. J. Evans: "Prognosis in acute disseminated encephalomyelitis; with a note on neuromyelitis optica." Quart. J. Med., 87:347-379, 1953.

Arus see in Infantile spasms: (salaam spasms)

EPILEPTIC SYNDROME

Syndromography

Clinic

This syndrome is made up of a group of disorders that usually begin in early childhood, but can appear at any age. It is characterized by recurrent and chronic paroxysmal alterations in neurological function, its common denominator being epileptic crises or attacks that manifest in a very varied way, from a brief " slip" in attention to a prolonged loss of consciousness accompanied by abnormal motor activity.

Epileptic seizures are classified according to the International Classification of Epileptic Seizures, based on the clinical manifestations of the seizure or the electroencephalographic pattern, both ictal and interictal, and its response to therapy.

Paraclinical examinations

Electroencephalogram. It has special value in the diagnosis of epilepsy, since it is capable of recording the electronic activity characteristic of the disease.

Sudden or spiked discharges, spike-slow wave complexes, and other abnormal paroxysmal hypersynchronous discharges are detected. It can also record focal anomalies typical of the lesion causing the epileptic focus.

However, the electroencephalogram has limitations, an isolated recording sometimes does not reveal the underlying epilepsy, therefore a normal electroencephalogram does not exclude the existence of epilepsy. In addition to the EEG at rest, it should be performed in other situations such as hyperventilation, light stimulation, and after sleep deprivation.

Syndromogenesis and etiology

The causes of epilepsy are many and several factors can coexist in the same patient.

It can be acquired as a result of a neurological or structural lesion of the brain or occur as part of many primary neurological diseases or present idiopathically, without a history of neurological injury or other obvious dysfunction.

Many types of metabolic abnormalities can cause the crisis.

In infants, the most common causes of seizures are anoxia or ischemia before or during childbirth, intracranial injury, metabolic disturbances, or congenital disorders of the development of the central nervous system.

In children, the most common cause is trauma and infections, but idiopathic crises are more evident.

Genetic factors can influence the development of this syndrome. Individuals with primary generalized seizures

(especially absence crises) present a marked family incidence. This hereditary mechanism is extremely complicated and the influence of multiple genes with variable penetrance is presumed.

In young children (6 months to 5 years), seizures are common during febrile illnesses, but it is unlikely that the child will develop epilepsy if the seizure lasts less than 5 minutes, is generalized, and does not present electroencephalogram or neurological alterations in the interictal period.

In adolescents and young adults, head trauma is the most frequent cause, as well as idiopathic or those related to the withdrawal of alcohol or medications that cause habits (drug addiction) and arteriovenous malformations.

In adults (30-50 years), brain tumors are the most common cause. From the age of 50, cerebrovascular diseases: embolism, hemorrhage, thrombosis (more rarely) or as a late sequel to these lesions, silent heart attacks and degenerative diseases.

Metabolic disorders are the cause of epilepsy that can occur at any age: hypoglycemia and hyperglycemia, hyponatremia and hypernatremia, hypocalcemia and hypercalcemia, uremia, and liver failure.

Most researchers propose that the common denominator of all epileptic seizures lies in the cerebral cortex, including the limbic cortex (hippocampus).

A rhythmic and repetitive hypersynchronous discharge of many neurons occurs in a localized area of the brain, which can be observed in the electroencephalogram. During epilectic discharges, metabolic phenomena occur that can contribute to the development of the focus, an increase in the extracellular concentration of potassium and a decrease in the concentration of calcium, which has an effect on neuronal excitation and the release of neurotransmitters and neuropeptides in abnormally large quantities that can act for a long time on central neurons and cause postconvulsive phenomena.

On the other hand, crises increase cerebral blood flow in the primarily affected areas, which causes an increase in glucose, alterations in oxidative metabolism and local pH.

Epileptic seizures are classified as:

1. Partial or focal seizures:
   a) Simple.
   b) Complex.
2. Partial or focal seizures with secondary generalization.
3. Generalized crises:
   a) Tonic-clonic seizures.
   b) Absence crisis.
   c) Bilateral myoclonus.
   d) Infantile spasms.
   e) Atonic crises.
) Tonic crises.
   g) Atypical absences.
4. State or status epilepticus:
   a) Tonic-clonic state.
   b) State of absence.
   c) Continuous partial epilepsy.
5. Recurrence patterns:
   a) Sporadic.
   b) Cyclical.
   c) Reflex (photomyoclonic, somatosensory, musicogenic, reading epilepsy).

1, PARTIAL 0 FOCAL CRISIS

They occur when neurons in a localized area of the cortex are activated, therefore the clinical manifestations will depend on the affected cortical area.

These crises are simple when they are not accompanied by alterations in consciousness or orientation, and complex when these alterations occur.

a) SIMPLE PARTIAL CRISIS

Syndromography

Clinic

They generally occur without loss of consciousness. Motor signs begin with tonic or clonic movements limited to one part of the body (the face and hands are the most frequent), producing recurrent muscle contractions caused by neuronal discharges in the corresponding area of the contralateral motor cortex. This muscle activity can spread from the affected area to adjacent ipsilateral parts of the body (from the right thumb to the right hand and from here to the arm and hemiface on the same side) producing the so-called " Jacksonian gait" in a matter of just 20-30 s. It may be accompanied by paresthesia, vertiginous sensations, auditory or visual hallucinations, unfounded feelings of fear, anger, delirium, complex hallucinations with discharges in the temporal and frontal lobes.

In some cases the unilateral motor signs are followed by turns of the head and eyes towards the side that is convulsing but sometimes towards the opposite.

Paraclinical examinations

Electroencephalogram. During crises, spike discharges are observed; during the inter-seizure period, irregular spike discharges may occur.

Syndromogenesis and etiology

The pathological process or focus of excitation usually resides in the Rolandic cortex (area 4) or motor cortex on the opposite side.

b) COMPLEX PARTIAL CRISIS

Synonymy

S. of temporal lobe epilepsy. S. of uncinate crisis.
S. of automation. Lennox psychomotor epilepsy.

Syndromography

Clinic

They appear at any age and are the most common of the chronic epileptic syndromes, characterized by episodic changes in the patient's behavior with loss of conscious contact with the environment that surrounds them.

They begin with auras of any type, often a complex hallucination or perceptual illusion: auditory, visual, olfactory or gustatory that patients come to relate as precursors to the crisis. Olfactory hallucinations allow this type of epilepsy to be diagnosed with great truthfulness.

During seizures, activity may be interrupted and only certain minor motor activity may be maintained (making noise with the fingers).

lips, swallowing, walking aimlessly), but they can be accompanied by the performance of unconscious activities that require great skill (driving a car, playing complicated pieces of music on the piano) and that last from a few seconds to minutes followed by confusion and amnesia, although many patients remember the aura. These symptoms are accompanied by ideational blockage and forced thinking and peculiar sensations of familiarity with events already seen, already thought about (dejá vu and déjá pensée).

Postictal anterograde amnesia and automatism are common. Aphasia may occur when seizures begin in the dominant hemisphere.

Paraclinical examinations

Electroencephalogram. It presents unilateral or bilateral spike or slow wave discharges over the temporal or frontotemporal regions between and during seizures. Exceptionally it is normal. Pharyngeal or sphenoid electrodes may record abnormal discharges, but in some cases only electrodes deep in the tonsillar nuclei or other limbic structures record seizure discharges.

Syndromogenesis and etiology

Seizures may be due to trauma during childbirth or the postnatal period, abscesses, tumors, heart attacks, vascular malformations or some other structural anomaly. They begin when neurons are activated in a localized area of the cortex in the temporal lobes, mainly the hippocampus or amygdala, or in any part of the limbic system, as well as in the parasagittal, mesial or frontoorbital region.

2. PARTIAL 0 FOCAL CRISIS WITH SECONDARY GENERALIZATION

Simple or complex partial seizures can evolve into generalized seizures with loss of consciousness and frequently accompanied by convulsive motor activity, which occurs immediately or within a few minutes. The focal origin of these generalized seizures is evidenced by the presence of an aura or the observation of any focal feature such as torsion of a limb, aphasia, and tonic deviation of the eye.

WIDESPREAD CRISIS

a) TONIC-CLONE CRISIS

Synonymy

Great evil crisis.

Syndromography

Clínica

It constitutes one of the most frequent types of epileptic seizures and an extremely unpleasant experience, both for the patient and for those accompanying him. It usually begins without prior warning, although sometimes the patient senses its possible imminence through a series of prodromes: he or she appears apathetic, depressed or irritated, or more rarely euphoric. This entire prodromal condition may be accompanied by abdominal pain or cramps, paleness or redness of the face, throbbing headache, constipation, or diarrhea.

There is a sudden loss of consciousness, tonic contraction of the muscles, loss of postural control, and the emission of a scream produced by the contraction of the respiratory muscles. rivers. The patient falls into opisthotonus and remains rigid for several seconds. There may be cyanosis as breathing is inhibited, followed by clonic, synchronous and recurrent contractions for 2 minutes, producing a sudden increase in blood pressure, elevation of body temperature, salivation and incontinence of the sphincters. Tongue biting may occur. At the end of the attack, the patient is left in a state of flaccidity and unconsciousness. Postictal depression can last from a few minutes to more rarely a day or more, gradually regaining consciousness but feeling exhausted with headache and desire to sleep. Some remain partially confused and do not regain functional normality for a few days.

Paraclinical examinations

Electroencephalogram. During tonic phase it shows rapid low-voltage activity (10 Hz) gradually turning into larger, slower waves across both hemispheres. In the clonic phase there are bursts of sharp waves related to rhythmic muscle contractions and slow waves that coincide with pauses. Between seizures the electroencephalogram is abnormal with polyspikes (or spikes) and waves, or sometimes sharp and slow wave discharges.

b) CRISIS OF ABSENCE

Synonymy

Small evil. Petit mal.

Syndromography

Clínic

They are characterized by their brevity and the lack of motor activity and sometimes the patients themselves are not aware of them. Seizures almost always begin in schoolchildren and young people (6-14 years old); they rarely occur after puberty. They tend to decrease in frequency towards adolescence and may disappear, but often give way to grand mal crises. There is a sudden interruption of conscious activity during which the patient remains motionless, fixes his gaze, stops talking and may be associated with clonic movements of the eyelids (eyelid flutter), facial muscles, and fingers or movements.

Synchronized movements of both arms.

When the absence is prolonged, automatism may occur that is confused with complex partial seizures. Postural tone is generally slightly decreased or increased and there is a slight vasomotor disorder. At the end of the crisis (2-10 s) the patient reestablishes contact with the environment and resumes his activity. There is usually no period of postictal confusion. Seizures can occur 100 or more times a day.

Paraclinical examinations

Electroencephalogram. Typical of this form of crisis. During them there are discharges of spikes and 3 Hz waves that appear synchronously in all leads.

Normal in inter-crisis periods.

c) BILATERAL MYOCLONI

Synonymy

Juvenile myoclonic epilepsy.

Syndromography

Clínic

They appear in adolescence. They can coexist with other types, but usually occur alone. They are characterized by sudden, brief, single or repetitive muscle contractions that affect a part of the body or the entire body.

Paraclinical examinations

Electroencephalogram. It shows polyspike discharges and slow sharp waves both within and between seizures.

d) INFANTILE SPASMS

Syndromography

Clinic

They begin in the first year of life in children with underlying neurological diseases, but also in supposedly healthy children and are characterized by brief synchronous contractions of the neck and torso, generally in flexion, accompanied by extension of the arms (spasms or tics of the salaam or greeting).

Paraclinical examinations

Electroencephalogram. There is a very disorganized background, high voltage slow waves that occur randomly, spikes and discharges and suppression (hypsarrhythmia patterns) that tend to disappear by 3-5 years to be replaced by other forms of generalized seizures.

e) ATONIC CRISIS

Syndromography

Clinic

They begin almost exclusively in childhood and are usually associated with diffuse brain lesions.

The ictal episode consists of brief loss of consciousness and postural tone of the patient, who simply falls to the ground without obvious cause.

Paraclinical examinations

Electroencephalogram. It has polyspigasys and slow waves.

) TONIC CRISIS

Syndromography

Clinic

They are less common and appear mainly in infants and children. They are characterized by sudden rigidity of the limbs or upper half of the body, frequently with deviation of the head and eyes to one side accompanied by a loss of consciousness that lasts 10-60 s. They are not followed by a clonic phase and are generally brief.

g) ATYPICAL ABSENCES

Syndromography

Clinic

They usually present in childhood, in those patients who have other forms of underlying neurological dysfunction.

They are similar to absence seizures, but can coexist with other forms of generalized seizures (tonic, myoclonic or atonic).

The most severe forms of this crisis (Lennox-Gastaut syndrome) present various types of generalized seizures frequently accompanied by mental deterioration.

Paraclinical examinations

Electroencephalogram. Shows spike and wave discharges at 2-4 Hz during seizures. During the interictal period it exhibits spikes or polyspike activities.

4. STATUS EPILEPTIC STATUS

Syndromography

Clínica

When prolonged or repetitive seizures occur without a recovery period between attacks, we are in the presence of a state or status epilepticus. It occurs in all forms of crisis.

If it is a tonic-clonic state, it constitutes a serious condition where the patient's life can be endangered.
In addition, there is physical exhaustion, which progresses to coma, tachycardia and dyspnea. An increase in temperature, circulatory collapse and nephrosis occur.

In absence seizures, the state of absence can be maintained for a long time.

In partial seizures, status epilepticus is called continuous partial epilepsy or focal motor status, where rhythmic clonic movements of one part of the body can persist for a variable period of time (from minutes to weeks or months) without spreading to other parts of the body.

Syndromogenesis and etiology

It occurs as a complication of epilepsy, grand mal or petit mal syndrome.

Literature

Grinker, R.R. and A. L. Sahs: Neurology. ed. 6, Springfield, Thomas, 1966.

Recurrence patterns

All recurrent seizures can be triggered sporadically or randomly, without having an obvious triggering phenomenon and can occur cyclically (sleep-wake cycle) or menstrually (catamenial epilepsy).

Reflex epilepsies due to reactions to a specific stimulus are less common. Among them we have:

Photomyoclonic or photoconvulsive: they are triggered by a light stimulus.

Musicogenic: triggered by a musical composition.

Somatosensory: 0occur due to tactile stimulation.

Reading or language: they are stimulated by reading and are characterized by brief myoclonic contractions of the jaw, cheek and tongue when the patient reads silently or in a low voice, which can evolve into generalized tonic-clonic seizures.

Literature

Aird, R. et al.: The epilepsies: A critical review. Raven Press, New York, 1984.

Browne, T. R. and R. G. Feldman: Epilepsy, diagnosis and management. Litele, Brown, Boston, 1983.

Delgado-Escueta, A.V. et al.: "Management of status apilepticus." N. Engl. J. Med. 306:1337, 1982.
Jasper, H. H. et al.: Basic mechanism of the epilepsy. Little, Brown, Boston, 1969.

Laidlaw, J. and A. Riche (eds.). A textbook of epilepsy, ed. 2d Churchill, Livingstone, London, 1982.
Penfield, W. and H. Jasper: Epilepsy and functional anatomy of the human brain. Little Brown, Boston, 1954.
Roger, J.; C. Drovet; M. Bureau et al.: Epileptic syndromes in infancy, childhood and adolescence. John Libbey Eurotext. Ltd, London, 1985.

Commission on classification and terminology of the ILAE.
Proposal for revised clinical and electroencephalographic classification of epileptic seizures. Epilepsy 22: 489, 1981.
Scot, A. K.: “Management of epilepsy." Br. Med. J., 288:986, 1984.

JACKSONIAN EPILEPSY SYNDROME

Synonymy

S. of focal seizures. S. de Bravais-Jackson. S. of cortical epilepsy.

Syndromography

Clínic

The seizure begins from a facial, brachial or crural focus that depends on the excited region of the motor cortex, and is frequently preceded by an aura of tremor; sometimes residual paresis remains.

Syndromogenesis and etiology

This syndrome is caused by organic diseases of the brain (trauma, abscesses, tumors, syphilis, etc.).

Literature

Jackson, J. H.: *Paralysis of tongue, palate and vocal cord." Lancet, 1:689-690, 1886.

KOJEWNIKOFF EPILEPSY SYNDROME

Synonymy

S. continuous partial epilepticus.

Syndromography

Clínic

It presents a condition characterized by high fever, delirium, rhythmic clonic jerks (extension or clonic flexion of a leg, elevation and descent of a shoulder, pronation and supination of the hand, grimacing with facial muscles, etc.), which can appear for weeks, months or years. During crises there is loss of consciousness.

Syndromogenesis and etiology

The syndrome is due to contralateral brain lesions, tumors, spring encephalitis and summer encephalitis, etc.

Bibliography

Kojewnikoff, A.Y.: "Osobyi: vid kortikalnoi epilepsi." Med. Obozr., 42:97-118, 1894.

ENDOCRINE EPILEPTIC SYNDROME

Synonymy

S. acromegalic epileptic.

Syndromography

Clínic

It appears in the first decade of life. It is characterized by epileptic seizures, myoclonic spasms and is accompanied by mental disorders and precocious puberty. There is accelerated growth and increase in body weight (acromegalic appearance in some cases).

Paraclinical examinations

Electroencephalogram. Patterns with deep subcortical lesions.

Syndromogenesis and etiology

The most common causes are: mammillary gland tumor or infundibular, trauma, inflammatory lesions with meningoencephalitis.

Literature

Bondin, G. and J. Barbizet: "D'association epilepsieendocrinopathie." Rev. Neurol., 91:330-347, 1954.

ERB SYNDROME

Synonymy

S. of spinal syphilis. S. of Erb-Charcot. S. de Strumpel. S. of spastic spinal paralysis. S. of Erb's spastic paraplegia.
S. of syphilitic amyotrophy.

Syndromography

Clínic

Erb syndrome appears in the tertiary period of syphilis, almost always 4-7 years after the chancre. It is more common in men. It begins slowly with sensations of heaviness and tingling in a lower limb accompanied by a syndrome of intermittent claudication of the spinal cord (Déjerine), with fatigue and weakness of the legs that forces the patient after a while on the road to drag his legs and even prevents walking. There is patellar and Achilles hyperreflexia with clonus, bilateral Babinski and urination disorders.

Paraclinical examinations

Lumbar puncture. Clear xanthochromic fluid with lymphocytosis, hyperalbuminosis and positive Pandy and Nonne reaction. Positive Wassermann reaction (frequently).

Syndromogenesis and etiology

Erb syndrome appears in the thetiary period of syphilis due to the action of Schaudinn's treponema on the central nervous system.

Literature

Erb, W. H.: Ueber syphilitische Spinalparalyse." Neurol. Centralbl. Leipiz., 11:161-168, 1892.

MULTIPLE SCLEROSIS SYNDROME

Synonymy

S. of polysclerosis. Sclerosis in plaques or insulae. S. of sclerosis disseminated in foci.

Syndromography

Clinic

It is a demyelinating condition of the central nervous system. It occurs almost exclusively in young individuals between 18 and 35 years old, it is rare in childhood. There are no sex differences, but women worsen significantly after pregnancy and constitute 60% of cases.

The presentation of the syndrome is varied and pleomorphic and the clinical picture is determined by the location of the demyelinating foci in the central nervous system.

The first attack can be declared by a single symptom or sign, which in 40% of cases is expressed by optic neuritis that is manifested by partial or total loss of visual acuity in one or both eyes, accompanied by pain during eye movements. Disturbances in color perception may indicate an early manifestation of the disease. Patients do not present with signs of papillitis or optic disc changes at onset. Diplopia due to injury to cranial nerves III, IV and VI or due to ophthalmoplegia due to injury to the medial longitudinal fasciculus, which is manifested by the inability to place one eye in adduction with the other in total abduction during lateral gaze. Multiple sclerosis may be accompanied by facial hyperesthesia or tic douloureux due to parity V; Paralysis or hemifacial spasms due to injury to the VII nerve, vertigo, vomiting and nystagmus (VII nerve) are also common, and deafness is less common.

Injury to cerebellar connections or spinocerebellar pathways causes ataxia with intentional tremor in the limbs and disorders of posture and gait.

Spinal cord lesions produce a great diversity of symptoms, interruption of the corticospinal or pyramidal tract causes the classic signs of motor neuron dysfunction (weakness-spasticity-hyperreflexia, clonus, Babinski sign and loss of abdominal skin reflexes).
There are disorders of conscious proprioceptive sensitivity due to seizure of the posterior cords (Burdach's Golly bundle).
Less frequently, thermal and painful disorders are present.

There are signs of bladder dysfunction: intermittent stream, frequency and incontinence, as well as intestinal function, particularly constipation. Men complain of sexual impotence, or dissociated genital potential: libido, erection and ejaculation without orgasm, although these alterations are more common.

rare

As the process evolves, the patient presents intellectual disorders with loss of attention, decreased memory, the most frequent emotional characteristic being depression due to the seizure of the subcortical and central white matter of the brain.

Local cerebral neurological signs (hemiparesis, homonymous hemianopsia and dysphasia) are rare, although they may be present.

Paraclinical examinations

Cerebrospinal fluid. It reveals only a slight increase in the number of cells (10 cells/mm'), predominantly T lymphocytes, although plasma cells may be found (rarely).

From the fluid of these patients it is possible to derive cell lines specifically reactive with various viral and non-viral antigens, which suggests that a heterogeneous immune response is being carried out.

90% of patients have less than 60 mg/dL of total proteins, values above 100 mg/dL should raise doubts about the diagnosis. Immunoglobulins G (IgG) are increased (the most characteristic feature), although at the beginning of the disease they may be normal.

Evoked potentials. Delay in the conduction of visual, auditory or somatosensory impulses (80 9% of patients).

Computerized axial tomography. It reveals low-density paraventricular or subcortical lesions, optic nerves, and brainstem. These lesions can be better evidenced if intravenous iodinated contrast solutions are used; Sometimes cortical atrophy is seen with widening of the ventricular cavities.

Nuclear magnetic resonance. It is more sensitive, it demonstrates the lowest density

In summary: elevated IgG in cerebrospinal fluid, abnormal evoked responses and images obtained by tomography and resonance are useful for the evaluation of the process, however, clinical data remain essential to establish the diagnosis.

Syndromogenesis and etiology

Immunological or infectious factors have been proposed to explain multiple sclerosis syndrome, but they are not conclusive.

The existence of an environmental factor (virus) perhaps geographically limited has been proposed to explain the development of the disease, however, all attempts to isolate, rescue or visualize the virus in these patients have failed.

The incidence in the black race is lower than in the white race, so genetic factors have been invoked. Blood relatives of patients (parents, siblings) are at greater risk.

Immune factors: the lesions of multiple sclerosis are similar to those of experimental allergic encephalitis (autoimmune disease induced in animals through immunization with myelin), in these cases the cause of the disease is the sensitivity of the lymphocytes Tauna basic protein of myelin, however, in multiple sclerosis it has not been possible to demonstrate sensitivity to this protein.

During the multiple sclerosis crisis, there is an increase in the secretion of prostaglandins by macrophages (which influence the properties of lymphocytes), causing a reduction in the function of suppressor cells, an increase in the number of activated T cells, an increase in the in vitro secretion of the dependent immunoglobulin from T cells, a decrease in the secretion of interferon and possibly a hypofunction of natural killer cells.

However, it is still unknown whether these changes have to do with the etiology of the disease.

It is suggested that there may be precipitating factors: infections, injuries or emotional disturbances, but they are not conclusive.

The etiology is unknown.

Literature

Cruveilhier, J.: Anatomie pathologique du corps humain, ou descriptions avec figures lithographiées et coloriées, des diverses alterations morbides dont le corps humain est susceptible.
Bailliere, vol. 2, Paris, 1829-1852.

MEDIAL LONGITUDINAL FASCICLE SYNDROME

Synonymy

S. of anterior internuclear ophthalmoplegia. St. of Lhermitte.

Syndromography

Clínica

Inability to adduct the eye beyond the midline or decreased adduction speed, accompanied by abduction nystagmus in the eye contralateral to the injury.

Syndromogenesis and etiology

The syndrome is caused by destruction of the medial longitudinal fasciculus at the level of the facial knee (pons).

The causes that cause it are: multiple sclerosis, gliomas of the pons, vascular disease, syphilis, alcoholism and inflammatory processes of the brain stem.

Literature

Bielschowsky, A.: "Die Innervation der Musculi recti interni als Seitenswaender". Sitzungsb. d. Ophth. Gesellsch., 164:171, 1903.

FAZIO-LONDE SYNDROME

Synonymy

Progressive bulbar palsy in childhood.

Syndromography

Clínica

Pediatric variety of spinal muscular atrophy that fundamentally affects the muscles innervated by the brain stem, characterized by facial dysplegia, dysarthria, dysphagia and dysphonia. It is sometimes accompanied by muscle weakness of the jaw and eye muscles, and rarely by progressive deafness.

Syndromogenesis and etiology

Hereditary motor neuron disease that is inherited as autosomal recessive, autosomal dominant, or sex-linked recessive.

Literature

Tandan, R. and W.G. Bradley: "Amyotrophic lateral sclerosis. Etiopathogenesis." Ann. Neurol., 18: 419, 1985.

CAROTID ARTERY-CAVERNOUS SINUS FISTULA SYNDROME

Syndromography

Clinic

Prodromes of headache and vertigo precede rupture (rare). Acute pain suddenly occurs in the eye and head; loss of consciousness is rare. Slowly progressive exophthalmos and a noise or shudder synchronous with the heartbeat that is perceptible by the patient, who describes it as a "buzzing" or "sawing noise."

Physical examination. Edema of the soft tissues of the orbit, dilation of the superficial veins of the eyelid and forehead, partial or total ophthalmoplegia of the affected eye. Compression of the caroid artery on the same side causes the noise to decrease or disappear. Fundus of the eye. Pulsation of the retinal veins, papilledemia, retinal edema, occasionally hemorrhages.

Paraclinical examinations

Radiology. Erosion of the sphenoidal walls of the orbit in advanced stages.

Arteriography. Shows the fistula.

Syndromogenesis and etiology

Spontaneous or traumatic arteriovenous fistula between the internal carotid and the cavernous sinus.

Literature

Hamby, W.B.: "Carotid-cavernous fistula; report of 32 surgically treated-cases and suggestions for definitive operation." J. Neurosurg., 21:859-866, 1964.

Travers, B.: “A case of aneurysm by anastomosis in the orbit, cured by ligation of common carotid artery." Med. Chir. Trans., 2:1-16, 1817.

FOIX SYNDROME

Synonymy

S. of the sphenoidal cleft. S. of the superior orbital fissure.

Syndromography

Clínic

Total ophthalmoplegia with eyelid ptosis and abolition of light and accommodation reflexes accompanied by anesthesia in the upper eyelid, forehead and root of the nose with abolition of the corneal reflex.

Syndromogenesis and etiology

This syndrome is caused by paralysis of the nerves that cross the superior orbital fissure on their way from the cranial cavity to the orbital cavity (pairs III, IV, VI, and ophthalmic branch of V).

It is caused by tumors in the vicinity of the sphenoid cleft (superior orbital fissure), internal carotid aneurysm, fractures of the lesser wing of the sphenoid, and syphilitic periostitis of the sphenoid cleft.

Literature

Brodal, A.: The cranial nerves in neurology anatomy in relation to clinical medicine. ed. 3. Oxford, New York. 1980. pp 448-457.

FRANKL-HOCHWART SYNDROME

Synonymy

Pineal-ophthalmic neurological S.

Syndromography

Clínica

Bilateral deafness, ataxia. Depending on the age of presentation, there are several manifestations of hypopituitarism. Limitation in looking upward. Constriction of the visual field.

Paraclinical examinations

Skull radiology.

Pneumoencephalography.

Cerebral angiography.

Computerized axial tomography of the skull.

Blood and urine. To demonstrate the presence of hypopituitarism.

Syndromogenesis and etiology

It is caused by a tumor of the pineal gland.

Literature

Geeraets, W. J.: Ocular syndromes. Lea & Febiger, Philadelphia, 1965.

Tassman, I: The eye manifestations of internal disease. ed. 3, Mosby, St. Louis, 1951.

Von Frankl-Hochwart, L.: "Ueber diagnose der Zirbeldrusentumoren." Deutsche Ztschr. F. Nervenh., 37:455, 1909;38:309, 1910.

GERSTMANN-STRÄUSLER-SCHEINKER SYNDROME

Syndromography

Clinic

It begins during the sixth decade of life, has a prolonged evolution (2-10 years), and is characterized by ataxia (spinocerebellar) and dementia. The signs and symptoms resemble olivopontocerebellar atrophy.

Syndromogenesis and etiology

It is considered a familial form of Creutzfeldt-Jakob syndrome, a familial degenerative condition that, despite having been shown to have an infectious etiology (infection by slow viruses due to unconventional agents), its causal agents have not been fully specified.

In the syndrome in question, degeneration of the anterior and posterior spinocerebellar and corticospinal tracts occurs.

This syndrome is coded in the McKusic catalog taking into account that the basis of the defect is due to a mutation of the prion protein gene.

Literature

Cecil: Textbook of Medicine. W.B. Saunders Co, Philadelphia, 1988.

Hudson, A. J. et al.: "Gerstmann-Sträussler-Scheinker disease with coincidental familial onset." Ann. Neurol., 14:670. 1983.

GODTFREDSEN SYNDROME

Synonymy

Ophthalmoneurological S. in nasopharyngeal tumors. S. neurological in cavernous sinus neoplasia.

Syndromography

Clinic

Ophthalmoplegia, more common in pair VI. Unilateral paralysis of the hypoglossal nerve and in most cases, anesthesia or neuralgia of the trigeminal nerve area.

Paraclinical examinations

Eye and neurological exams.

Syndromogenesis and etiology

Malignant nasopharyngeal tumor that takes over the cavernous sinus and metastasizes to the retropharyngeal lymph nodes, compressing the nerves of pair XII, never in the hypoglossal canal.

Literature

Godtfredsen, E.: "Ophthalmoneurological symptoms in malignant nasopharyngeal tumors." Br. J. Ophth., 31:78-100, 1947.

GILLES DE LA TOURETTE SYNDROME

Synonymy

S. of generalized tics with coprolalia. S. of impulsive myospasia. S. de Brissaud II. S. de Tourette. Maladies des tics convulsifs. S. of psychogenic tic. S. of painless or non-painful tic. Gilles de la Tourette's disease.

Syndromography

Clínica

This syndrome appears in childhood between the ages of 7 and 15, and is characterized by a convulsive movement that is usually located preferentially in the mimic muscles, more rarely in the trunk and limbs, which increases with emotions and fatigue and decreases with distractions or during moderate physical exercise and disappears during sleep.

The movements or series of movements are sudden, recurring and brief. They are usually located on the face, and rarely on the trunk and limbs. The onset of puberty accentuates them. Patients complain that they cannot avoid them. Over time, the patient begins to quietly exclaim words or short phrases of obscene meaning (coprolalia).

Echolalia, polylalia, and ecomimia may also occur.

There is hyperactivity and attention disorders. Sometimes states of obsession and delirium occur that can lead to dementia.

Syndromogenesis and etiology

Tics can be of two types: psychogenic or functional (more frequent) and organic, resulting from injuries to the extrapyramidal system, especially the striatum.

The psychogenic ones appear in childhood, the organic ones in old age.

Observation in other family members has provided evidence that the disease is hereditary, but its transmission pattern has not been proven, although autosomal recessive inheritance is possible in some families.

The etiology is unknown.

Literature

Gilles de la Tourette, C.E.A.B.: "Etude sur une affection nerveuse caractérisée par l'incoordination motrice, accompagnée d'echolalie et de coprolalie." Arch. Neurol., 9: 19-42e 158-200.
Paris 1885.
Trousseau, A.: Clinique Médicale de l'Hotel-Dieu de Paris. vol. i.
1873, p.855.

GUNN SYNDROME (MARCUS)

Synonymy

S. of blinking-mandibular.

Syndromography

Clínica

It presents from birth, with moderate ptosis of one of the eyelids when the mouth is opened. The mandible has a lateral deviation, towards the opposite side of the ptosis, resulting in elevation of the upper eyelid with opening of the palpebral fissure. When acquired it can appear at any age.

Syndromogenesis and etiology

It can be congenital or in some cases acquired.
The etiology is unknown.

Literature

Gunn, R. M.: "Congenital ptosis with peculiar associated movements of the affected lid." Tr. Ophthal. Soc. U. Kingdom, 3:283-286. 1883.

Mohan, H.: A. N. Gupta and S. K. Jain: "Marcus Gunn phenomenon with pardoxical oculo-palpebral movements." Br. J. Ophthal., 51:780-782, 1967.

Wartenberg, R.: "Winking-jaw phenomenon." Arch. Neurol. Psychiat., 59:734-753, 1948.

HALLERVORDEN-SPATZ SYNDROME

Synonymy

S. of pigmentary degeneration of the globus pallidus. C. and Vogt's pallidum status desmyelinisatus. S. of state pigment-
Helfand state.

Syndromnography

Clinic

This syndrome appears approximately between 7-10 years of age. It is more common in males. It presents with a gradual increase in rigidity of all limbs, starting with the lower ones, inversion of the feet (pes cavus), dystonic postures, tremor, athetosis, sometimes there is hyperreflexia and Babinski's sign. There is dysarthria (monotonous and unintelligible language), the facies adopts a fixed expression. There are intellectual disorders that lead to dementia. Retinitis pigmentosa (sometimes). Optic atrophy has been described.

Paraclinical examinations

There is no biochemical test that corroborates the diagnosis. No abnormalities have been found in the serum concentration of iron or its metabolites.

Syndromogenesis and etiology

The dopamine-neurome-lanin system may be involved in the pathogenesis of this syndrome.

It has a genetic etiology with autosomal recessive inheritance. The basic defect is unknown.

Literature

Elejalde, B. R.; M. M. Elejalde and F. López: "Hallervorden-Spatz disease." Clin. Genet, 16:1-18, 1979.
Hallervorden, J.: Die degenerative diffuse sklerose. In Lubarsch,
EITHER.; F. Henke, and R. Rossie, (eds.): Handsbuch der speciellen pathologischen Anatomie und Histologie. Berlin, Springer- -Verlag, vol. 13, 1957, pp. 716-782.

ACUTE SUBDURAL HEMATOMA SYNDROME

Syndromography

Clinic

The syndrome is established after a head trauma, sometimes with temporary loss of consciousness and is manifested by: headache, drowsiness, agitation, mental slowness and confusion that progressively worsen, until reaching coma.
Physical examination. The main signs are: unilateral dilated pupil, loss of corneal reflex, congestion of the veins in the fundus of the eye, progressive hemiparesis of the arm and leg contralateral to the location of the hematoma.

The prognosis is poor with high mortality, which reaches 75-90% even when the clot is evacuated. Early diagnosis and surgical intervention are essential.

Paraclinical examinations

Computerized axial tomography. It can demonstrate the injury in most cases.

Arteriography. It demonstrates the deviation of the central structures due to the compression of the hematoma that behaves like a brain tumor.

Syndromogenesis and etiology

The syndrome is caused by a head trauma that causes blood accumulation in the subdural space due to tearing or laceration of the cortical veins on their way to the superior longitudinal sinus.

Literature

Grinker, R. R. and A. L. Sahs: Neurology, ed. 6, Thomas, Springfield, 1966.

Jennett, B. and G. Teasdale: Management of head injuries. Davis, Philadelphia, 1985.
Miller, A. D.: “Acute traumatic midbrain hemorrhages.” Ann. Neurol., 18:80, 1985.
Wilkins, R. H. and S. S. Reganchary (eds.). Neurosurgery (vol.2) New York, McGraw Hill, 1985. pp:1531-1693.

CHRONIC SUBDURAL HEMATOMA SYNDROME

Syndromography

Clinic

This syndrome occurs in children and adults, especially in alcoholics. Symptoms begin weeks or months after a moderate or severe head trauma. It's about progression

gradual. It is characterized by severe persistent headache, some patients present seizures as the first symptom, irritability, apathy, sleep attacks, lethargy and. finally, stupor and coma.

Physical examination. Hemiplegia, usually contralateral, sometimes ipsilateral. Eventually patients may present papilledema, external ophthalmoplegia and mydriasis, rarely hemianopia; Sensory changes are not frequent.

Paraclinical examinations

Lumbar puncture. Cerebrospinal fluid pressure is normal, clear or slightly xanthochromic in appearance, protein level and cell count are normal or slightly elevated.

two.

Skull radiology. Negative, but displacement of the pineal body may be observed if it is calcified, in case of herniation due to compression of the brain tissue.

Electroencephalogram. It may show area of cortical activity in the area of the lesion.

Angiography. The arterial branches appear separated from the skull. The middle cerebral artery and the anterior cerebral artery may be displaced to the opposite side.

Echoencephalogram

Computed axial tomography and nuclear magnetic resonance. They constitute the diagnostic methods of choice. They show a low-density mass on the convexity of the hemisphere, but sometimes only a deviation of the midline structures and compression of the lateral ventricles is observed, because the clot becomes isodense with the adjacent brain, after 2-6 weeks.

Brain scan with radionuclides in anterior projection. It is often the most useful method to confirm the diagnosis.

Exploratory bilateral trepanation. It should be used when the clinical picture has not been diagnosed.

Syndromogenesis and etiology

This syndrome is due to head trauma. In reality, they are not true hematomas, but hemorrhagic pseudocysts that, as they disintegrate, increase osmotic pressure and trap fluid, which progressively increases the volume of the cyst.

Especially in elderly people, the clinical picture may be preceded by a trivial blow that may even have been completely forgotten.

Literature

Langfit, T.W. andT. A. Genarelli: "Can outcome from head injury be improved?" J Neurosurgery, 56:19, 1982.
Martuza, R. L. and T. M. Aquino: Trauma. In Samuels, M. A. (ed.) Manual of neurological therapeutics. Little, Brown, Boston, 1986

SUBDURAL HEMATOMA SYNDROME IN CHILDREN

Syndromography

Clínic

It occurs during the first six months of life and is characterized by vomiting, loss of appetite, irritability, seizures and stupor, and low-grade fever.

Physical examination. Bulging of the fontanels, elongation of the skull, distension of the scalp veins, retinal hemorrhages and rarely papilledema.

Paraclinical examinations

Subdural space puncture. It should be performed through the lateral angle of the anterior fontanelle or in the coronal suture.

Syndromogenesis and etiology

This syndrome is due to head trauma during childbirth or in the postnatal period.

Literature

Grinker, R. R., and A. L. Sahs: Neurology, ed. 6, Thomas, Springfield, 1966.

SUBACUTE SUBDURAL HEMATOMA SYNDROME

Syndromography

Clinic

It appears a week, or later, after a head trauma. There is headache, irritability, neck stiffness, hemiplegia and aphasia.

Physical examination. Hemiplegia with Babinski sign, bradycardia, alteration in respiratory rhythm and strabismus is found. Mortality is 25%.

Paraclinical examinations

Skull radiology.
Exploratory trepanation.
Angiography.
Computed axial tomography.

Syndromogenesis and etiology

Acute subdural hematoma is due to cranial trauma, which causes a tear or laceration of the brain tissue and/or the satellite veins on their way to the superior longitudinal sinus.

Literature

Grinker, R. R. and A. L. Sahs: Neurology, ed. 6, Thomas, Springfield, 1966.

TRANSIENT POSICTAL HEMIANOPSIA SYNDROME

Syndromography

Clínica

The syndrome is manifested by homonymous hemianopia with or without visual hallucinations, which disappears after a few days and may return with or without new apoplectic stroke attacks.

Syndromogenesis and etiology

It is caused by hypoxia of an area of the brain that was previously damaged.

Literature

Walsh, F. B.: Clinical Neurolophthalmology. ed. 2, Williams & Wilkins, Baltimore. 1957.

SYNDROME OF ALTERNATING HEMIPLEGUS WITH HYPOGLOSSUS INJURY

Synonymy

S. de Déjerine. S. interolivary. S. de Reynold-Revilliod-Déjerine. S. paramedian of Foix.

Syndromnography

Clinic

Clinical picture characterized by controllateral hemiplegia, which spares the face and lingual paralysis with ipsilateral peripheral hemiatrophy and fibrillation. Sometimes ipsilateral tactile hemianesthesia.

Syndromogenesis and etiology

Produced by vascular or tumor lesions that affect the pyramidal pathway and fibers of the XII cranial nerve (hypoglossal nerve).

ALTERNATE HEMIPLOGY SYNDROME

Synonymy

S. of hypoglossic alternating hemianesthetic hemiplegia.

Syndromography

Clinic

This syndrome is manifested by ipsilateral paralysis and hemiatrophy of the tongue (due to injury to the nucleus or already crossed fibers emerging from the hypoglossal nerve). Contralateral hemiplegia that spares the face (due to involvement of the pyramidal fibers not yet crossed) and loss of vibration sensitivity (palesthesia) and position (bathisesthesia) on the same side as the hemiplegia, due to seizure of the medial lemniscus without loss of thermal and pain sensitivity.

Syndromogenesis and etiology

The syndrome is produced by injury to the anterior and middle regions of the medulla oblongata that involves the spinal tract and the emergence of the fibers of the hypoglossal nerve, as well as the medial lemniscus.

Literature

Alpers, B.J.: Clinical Neurology, ed. 5, Davis, Philadelphia, 1963.

EXTRADURAL OEPIDURAL HEMORRHAGE SYNDROME

Syndromography

Clínica

This syndrome appears in patients with a history of head trauma with temporary loss of consciousness, who after an interval (several days or weeks), begin to present headache of progressive intensity, vomiting, drowsiness, confusion, epileptic contractions (unilateral) and hemiparesis.

Physical examination. Increased tendon reflexes and Babinski sign. Breathing becomes deep and stertorous; later, superficial and irregular until reaching respiratory arrest. There is bradycardia, narrow pulse, elevation of systolic pressure. The pupil is dilated on the side of the hematoma.

Paraclinical examinations

Cerebrospinal fluid. Increased tension (sometimes normal). Clear or bloody (due to associated subarachnoid hemorrhage).

Skull radiology. Visualization of the fracture line through the groove of the middle meningeal artery.

Computerized axial tomography and carotid arteriography. They allow you to confirm the location.

Syndromogenesis and etiology

It is due to hemorrhage in the space between the dura mater and the diploe, generally caused by the rupture of the middle meningeal artery due to a fracture of the temporal or parietal bone, or by a laceration of the venous sinuses of the dura mater (less common), compression of the cerebral hemispheres and sometimes cerebral infarction.

Literature

Heyser. J. and G Weber: "Die epideuralen Hamatome." Schweiz. Med. Wschr., 94:2-7; 46-52, 1964.

HERTWIG-MAGENDIE SYNDROME

Synonymy

S. of gaze deviation. Hertwig-Magendie phenomenon.

Syndromography

Clinic

This syndrome is characterized by vertical diplopia (one eyeball deviates downward and the other deviates up and out).

Syndromogenesis and etiology

It is due to trauma (spontaneous or surgical), atherosclerosis, neoplastic lesion of the posterior cranial fossa, vascular lesions of the brain stem. It can also be caused by syringobulbia with demyelinating lesions.

Literature

Hertwig, H.: Experimenta quaedam de effectibus et de verosimiliharum partium functione, Berolini, formis Feisterianis et Eiserdorffianis (1926) . Ind. Cat. Surg. Gen., Ist. series, 6:185, 1885.

SUBDURAL HYGROMA SYNDROME

Syndromography

Clinic

Symptoms and signs are similar to those of subdural hematoma syndromes.

Syndromogenesis and etiology

The etiology is due to head trauma. As a result of the trauma, a collection of clear or yellowish fluid occurs in the subdural space that has a high concentration of proteins.

Literature

Naffziger, H. C.: "Subdural fluid accumulation following head injury." J.A. M.A., 82:1751-1752, 1924.
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ENDOCRANIAL HYPERTENSION SYNDROME

Syndromography

Clinic

It is characterized by a classic triad:

1. Headache. It has a particular character. It begins paroxystically and later continuously. It is intense, persistent, is accompanied by a feeling of weight (gravity) and is not relieved with common analgesics, it worsens with cubitus.
   It is relieved by standing or sitting and is exaggerated under any circumstance that varies the tension of the cerebrospinal fluid (coughing, sneezing, defecation, etc.). The headache can extend to the entire head or be localized (frontal, temporal, parietal, etc.).

2. Vomiting. They are produced by the compression of the medulla oblongata and excitation of its centers. They are spontaneous, not preceded by nausea, in bursts (cerebral vomiting), although this is not always the case, they can adopt other modalities and present the same characteristics as in gastric conditions.

3. Papillary edema. It constitutes a sign of capital importance.
   It occurs in 80% of patients, and is translated by visual disorders (floaters, amblyopia and intermittent blindness).

These three fundamental symptoms can be accompanied by other manifestations such as:

1. Bradycardia (due to central action on the parasympathetic nucleus of the vagus), which is sinus type (50-60 beats/min).
   Slow and progressive intracranial hypertension does not modify the pulse.

2. Increased blood pressure (it does not rise in slow and progressive hypertension).
3. Bradypnea, with increased amplitude that can reach Cheyne-Stokes breathing in cases of rapidly establishing hypertension.

Vertigo, generalized epileptiform seizures, transient ataxia, constipation, drowsiness and psychological alterations are not uncommon.

Physical examination. Fundus of the eye. Venous congestion is observed with enlarged veins and narrowing of the retinal arteries, perivenous hemorrhages may appear. Blurred and raised edges of the papilla, with disappearance of the central excavation or fovea of the papilla, producing enlargement of the blind spot and concentric narrowing of the visual field with, therefore, a decrease in visual acuity that evolves towards atrophy of the papilla and total loss of vision.

In children, separation of the skull sutures is observed.

Areflexia, superficial and deep, accompanied by signs and symptoms depending on the etiology of the syndrome, which vary according to its location and nature.

Paraclinical examinations

Lumbar puncture. It must be practiced with extreme caution (do not let the liquid flow freely). Increase in pressure (greater than 200 nm of water).

Electroencephalogram. If hypertension is marked, there may be a general slowing of the waves.

Computed axial tomography and nuclear magnetic resonance. They allow detecting tumors or ventricular deformities.

Carotid arteriography. It detects aneurysms of the arterial circle of the brain and in the case of tumors, the displacement of vessels and alterations of the circle of Willis, or hypervascularized areas.

An avascular zone is observed in brain abscesses.

Syndromogenesis and etiology

Intracranial pressure can increase due to a reduction in the capacity of its continent, as occurs in the collapse of the bones of the skull, skull tumors that develop inwards, disorders of bone development that cause premature closure of the sutures of the skull, or due to an increase in its content, as in brain tumors, vascular malformations, brain abscesses, meningoencephalitis and malformations. congenital mations.

Cerebral edema is the most important factor in the production of this syndrome. By increasing the content of the cranial cavity in the course of intracranial tumors, abscesses, trauma and meningoencephalitis, there is compression of the nervous tissue, venous collapse, which causes difficulty in venous drainage and consequently edema: on the other hand, there is passive congestion of the venous plexuses, increased production of cerebrospinal fluid and decreased reabsorption.

Cerebrospinal fluid circulation disorders are also a very important factor in the production of this syndrome. When there is an obstacle to the free flow of the fluid, an accumulation of fluid occurs above it, which causes intracranial hypertension due to hydrocephalus, as observed in the tumors themselves that in their growth displace and obstruct the fluid circulation pathways, or during inflammatory processes that seal the communication orifices of the ventricular system (foramen of Monro, Magendie or Luschka); During trauma, these pathways can be obstructed by the presence of blood clots formed in the ventricular cavities.

Blood circulation disorders can lead to intracranial hypertension. Any obstacle to venous drainage from the cranial cavity, as occurs in the course of tumors of the mediastinum, lung and to a lesser extent in right heart failure, causes venous congestion, difficulty in the reabsorption of cerebrospinal fluid and consequently its increase.

In the course of meningoencephalitis, a disorder in the production of cerebrospinal fluid may also occur, due to irritation of the caroid plexuses of the ventricular cavities.

In subarachnoid hemorrhage, great blood loss occurs in this space, which due to its rapidity exceeds the physiological possibilities of reabsorption, causing intracranial hypertension.

Literature

Welsberg, L. A.: "Bening intracranial hypertension." Medicine (Batilmore), 54:197, 1978.

BENIGN ENDOCRANEAL HYPERTENSION SYNDROME

Synonymy

S. of pseudotumor cerebri. S. de Nonne.

Syndromography

Clinic

It is seen more frequently in obese adolescents. It is characterized by headache, papilloedema, paralysis of the VI nerve (n. abductor), causing a convergent strabismus and progressive loss of vision.

Paraclinical examinations

Computerized axial tomography. Decrease of the lateral ventricles in the form of a slit.

Lumbar puncture. Increased cerebrospinal fluid pressure with decreased protein.

Syndromogenesis and etiology

Generally, the cause is unknown, although it is associated with pregnancy, administration of corticosteroids, use of sex hormones, disorders of adrenal and parathyroid origin, venous sinus thrombosis, consumption of vitamin A, tetracycline or nalidixic acid.

Literature

Wisbertg, L. A.: "Benign intracranial hypertension." Medicine (Batilmore), 54:197, 1978.

HIRANO-PARKINSONISM SYNDROME

-DEMENTIA

Syndromography

Clinic

This syndrome is observed exclusively among members of the Chamorro race on the island of Guam. It appears between 32-64 years with an average of 52 years, predominates in the male sex, has an insidious appearance and is not preceded by phallitis or other febrile illness.

It is characterized by akinesia, facial inexpression, poor coordination of alternating movements, mental deterioration, mild tremor, moderate rigidity, and generalized hyperreflexia. In some cases it may be accompanied by signs and symptoms of sclero-amyotrophic lateral syssis.

Syndromogenesis and etiology

This syndrome can be considered as an entity that represents the combination of parkinsonism, presenile dementia and amyotrophic lateral sclerosis, without, however, having all the characteristics of each of them. The etiology is unknown.

Literature

Hirano, A.; L.T. Kurland; R. S. Krooth et al.: "Parkinsonism- dementia complex, endemic disease on island of Guam. J. Clinical features." Brain, 84:642-661, 1961.

HOLMES I SYNDROME

Synonymy

S. of visual disorientation. S. of disordered spatial perception.

Syndromography

Clinic

Holmes I syndrome is characterized by a lack of ability to locate moving or stationary objects in all three spatial dimensions and by a loss of the ability to estimate distance and adequately judge the size and length of objects.

Physical examination. Visual acuity is preserved, difficulty maintaining gaze fixation when the object is moving, visual agnosia, failure to accommodate and convergence, and absence of eyelid reflex.

Syndromogenesis and etiology

It may be due to bilateral brain injury to the posterior part of the parietal lobe or surface of the occipital lobe, caused by trauma, hemorrhage, etc.

Literature

Riddoch, G.: "Visual disorientation in homonymous half fields." Brain, 58:376-382, 1935.

HOLMES II SYNDROME

Synonymy

S. of primary parenchymal degeneration of the cerebellum. S. of olivocerebellar atrophy. S. of familial cerebellar cortical atrophy.

Syndromography

Clinic

It is rare in appearance, starting around the age of 30. It begins with a staggering drunken gait followed by manual clumsiness, motility disorders of the lower limbs, dysarthria (slow and hesitant speech), intention tremor and nystagmus.

Syndromogenesis and etiology

There is degeneration of the cerebellum with atrophy of the inferior olivary nucleus and olivocerebellar connections. The middle cerebellar peduncle and spinal cord are not affected. The etiology is unknown.

Literature

Holmes, G.: "A form of familial degeneration of the cerebellum." Brain, 30:468-489, 1907.

STIFF MAN SYNDROME

Synonymy

S. de Moersch-Woltmann. S. of progressive muscle rigidity. S. of fusiform myotonia.

Syndromography

Clinic

It begins in mid-adult life, affecting the male sex more frequently.

It begins with intermittent stiffness and spasms of the muscles of the trunk and limbs that become more and more continuous until affecting the muscles of mimicry, swallowing and finally the respiratory muscles, with no other abnormalities of the nervous or muscular system detected.

There are frequently paroxysmal painful tetanic spasms, provoked by emotional and/or physical stimuli that can cause bone fractures. These spasms disappear during sleep. There are no sensory or intellectual disorders. Profuse sweating and tachycardia may appear.

Paraclinical examinations

Urine. In 30% of cases there is glycosuria.

Syndromogenesis and etiology

Spinal cord disinhibition of unknown nature. The loss of inhibitory (input) neuronal impulses to the cells of the anterior horns can produce the repeated discharge of motor neurons and cause painful and very intense muscle contraction. Its relationship with hyperthyroidism and/or hypothalamic disorders has been assessed.

The etiology is unknown although affected families have been reported, which suggests an autosomal dominant inheritance.

Literature

Adams, R. P.: Diseases of muscle. A study in Pathology. ed. 3 Harper and Row, New York, 1975.

Bartholow, R.: *Aneurysms of the arteries at the base of the brain, their symptomatology, diagnosis, and treatment." Amer. J. Med. Sci., 64:373-386, 1872.

Miller, E and H. Korsvik: "Baclofen in the treatment of the stiff-man syndrome." Ann. Neurol., 9:511.1981.

HOMEN SYNDROME

Synonymy

Post-concussion S. S. of punch-drunk. S. of the boxer. S. of pugilists' encephalopathy.

Syndromography

Clínic

This syndrome appears in boxers after a certain number of years in the ring. The patient presents with forgetful memory, slowing of thinking and dysarthria, accompanied by slow, coarse and imprecise movements, unstable gait with an increase in the base of support.

Paraclinical examinations

Sometimes there is deafness and loss of visual acuity, loss of facial expression, which is similar to that of parkinsonians.

Electroencephalogram. Slow waves of theta type and sometimes delta type.

Pneumoencephalogram. Dilatation of the lateral ventricles and an excavated septum pellucidum that distinguishes them from other forms of cerebral atrophy.

Syndromogenesis and etiology

This brain injury occurs as a result of the cumulative effect of repeated brain trauma, and is observed almost exclusively in professional boxers.

Literature

Homen, E.A.: Eine eigenthumliche Familien Krankheit unter der Form progressiven Dementia, mit besanteren anatomischen. Befund. Neurol. Zbb., 9:514-518, 1890.

HUNT I SYNDROME (RAMSAY)

Synonymy

S. of auricular herpes zoster. S. of geniculate neuralgia. S. of herpes zoster of the geniculate ganglion. S. by Ramsay Hunt.

Syndromography

Clinic

Hunt syndrome I occurs at any age, more common in the elderly, and is characterized by an erythematous maculopapular rash, which rapidly evolves to form vesicles and is accompanied by intense unilateral zonatous pain in the corresponding dermatome (external auditory canal and external acoustic meatus, tympanic membrane and pinna).

It is a unilateral herpetiform vesicular eruption that affects branches of the trigeminal nerve. The lesions appear on the face, mouth, tongue and eyes, as well as on the auditory nerve, also affecting the geniculate ganglion and the sensory branch.

sorial facial nerve.

Physical examination. Alterations of taste and sensitivity in the anterior two-thirds of the corresponding half of the tongue (ageusia and hypoesthesia), due to paralytic irritation of the afferent taste fibers of the geniculate ganglion, coming from the chorda tympani (facial nerve). Concomitant ipsilateral facial paralysis, which occurs on the second or third day after the zone. Sometimes a variety of this Zone can be observed in which auditory disorders are associated: tinnitus, hearing loss and Meniere-type vertigo due to propagation to the vestibulocochlear nerve.

Paraclinical examinations

Tzanck smear. It is carried out by scraping the base of the lesions and demonstrates the presence of multinucleated giant cells.

Antibodies. Tests can be performed: against membrane antigens (FAMA), immune adhesion hemagglutination, or the enzyme immunosorbent assay (ELISA), which are quite specific.

These tests are not done in routine practice, but some specialized centers can isolate the virus in cell lines in vitro, or demonstrate seroconversion or increase in the varicella zoster virus antibody titer.

Syndromogenesis and etiology

The syndrome is caused by a viral infection caused by the reactivation of the varicella virus, which remains latent in the posterior root ganglia after chickenpox. Its exact pathogenesis is not well known, but it has been postulated that the virus spreads centripetally through the sensory nerves to the sensory ganglia, where it remains in a latent state associated in some way with neurons and astrocytes.

Viral replication is then activated, in most cases without obvious triggering cause. It is thought that the decrease in immunity due to age and lack of exposure to children with chickenpox causes the infection to reactivate. Once

Activated, the virus multiplies in the ganglia (ganglionitis), causing pain along the sensory distribution, multiplying again in the skin and causing the appearance of vesicles.

Literature

Brunell, P. A. et al.: "Prevention of varicella by zoster immune globulin." N. Engl. J. Med., 280:1191, 1969.
Hope-Simpson, R. E.: "The nature of herpes zoster: A long-term study and a new hypothesis." Proc. Royal Soc. Med., 58:9, 1965.
Hunt, J. R.: "On herpetic inflammations of geniculate ganglion: a new syndrome and its aural complications." Arch. Otol., 36:371-381, 1907.

HUNT II SYNDROME (RAMSAY)

Synonymy

S. of myoclonic cerebellar dyssynergia.

Syndromography

Clinic

The average age of onset is young adulthood or childhood. It is characterized by myoclonic seizures that begin locally in one of the limbs and subsequently extend to the entire skeletal muscular system. The lower limbs are less affected than the upper ones. There is asthenia, dysarthria, dysmetria, hypotonia, adiadochokinesia. Mental deterioration is rare.

Syndromogenesis and etiology

There is an almost complete loss of the cells of the dentate nucleus, as well as atrophy of the columns of Goll and the anterior and posterior spinocerebellar tracts.

It is inherited in an autosomal recessive manner.

The etiology is unknown.

Literature

Hunt, J.: "Dyssynergia cereballaris myoclonica-primary atrophy of the dentate system, a contribution to the pathology and symptomatology of cerebellum." Brain, 44:490-538, 1921.
May, D. L. and H. H. White: Familial myoclonus, cerebellar ataxia and deafness." Arch. Neurol., 19:331-338, 1968.

TRANSIENT CEREBRAL ISCHEMIA SYNDROME

Synonymy

S. of intermittent cerebrovascular insufficiency. S. of threatening stroke. S. of intermittent cerebrovascular claudication. S. of incipient stroke.

Syndromography

Clinic

This syndrome is characterized by its sudden onset and cessation, sometimes gradually but frequently abruptly, whose duration varies between seconds and hours. It is sometimes triggered when the patient moves from the clinostatic or sitting position to the upright position, or when the head rotates (usually there is no triggering factor). The symptoms are related to the affected vascular system.

When the carotid system is affected, the patient presents episodes of hemiparesis, contralateral hemianesthesia and amblyopia or amaurosis on the same side as the ischemia.

On rare occasions it may be accompanied by headaches, tonic-clonic seizures without loss of consciousness.

If the lesion occurs at the level of the vertebrobasilar system, the clinical picture consists mainly of vertigo, paresis or dysesthesia of one hemibody, monoparesis or tetraparesis, diplopia, dysarthria, facial or perioral numbness, dysphagia, unilateral or bilateral homonymous hemianopsia, occipital headache and more rarely hiccups, drowsiness and obtundation of the head. consciousness. All symptoms do not usually occur in the same patient, but rather one or two occur, forming a varied symptom complex.

Syndromogenesis and etiology

It is due to a decrease in arterial flow, which produces a local reduction in perfusion pressure that deprives brain tissue of oxygen and other metabolites.

The most frequent causes are: atherothrombosis of intracranial or extracranial arteries, embolism of cardiac origin, pulmonary embolism, cardiovascular diseases that reduce cerebral blood flow, lacunar infarction, generalized cerebral hypoxia, non-atherosclerotic vasculopathy, cerebral arterial spasm, thrombosis of the cerebral sinuses and veins.

JACKSON SYNDROME

Synonymy

S. vagoaccessoryhipogos. Hughlings-Jackson S.

Syndromography

Clínica

This syndrome is manifested by velopalatine and laryngeal paralysis and paralysis of the sternocleidomastoid, trapezius and larynx, accompanied by paralysis and atrophy of the peripheral tongue. There is contralateral hemiplegia.

Syndromogenesis and etiology

The causes that most frequently produce this syndrome are: acute anterior poliomyelitis, progressive bulbar or labioglossopharyngeal paralysis. It is also seen in syringomyelia and amyotrophic lateral sclerosis.

These conditions cause paralysis of the pneumogastric, spinal, and hypoglossal muscles, accompanied by injury to the not yet decussated pyramidal tract.

Literature

Jackson, J.H. : *Paralysis of tongue, palate, and vocal cord.'" Lancet, 1:689-690, 1886.

JACOD SYNDROME

Synonymy

S. of the petrosphenoidal carrefour. S. de Negri-Jacod.

Syndromography

Clínica

It is manifested by unilateral trigeminal neuralgia, lesion of the optic tract (unilateral amaurosis), unilateral total ophthalmoplegia.

Hearing loss (middle ear) and unilateral or bilateral cervical lymphadenopathy (30% of cases), paralysis of the velopalatine muscles.

Syndromogenesis and etiology

The lesions that most frequently cause this syndrome are: primary neoplasms or metastasis, sarcoma of the Eustachian tube, skull base, or pituitary gland.

The petrosphenoidal fossa forms an internal vertex angle limited in front, by the greater wing of the sphenoid, and behind, by the temporal pyramid. Said angle includes:
the middle cranial fossa, where the maxillary (foramen rotund) and mandibular (foramen ovale) branches of the trigeminal nerve emerge, and the cavernous sinus, where cranial nerves II, IV and VI pass, accompanied by the ophthalmic branch of the trigeminal nerve.

Tumors that settle in this region cause paralysis of the optic nerve, as well as cranial nerves III, IV, VI, and neuralgia and hyperesthesia of the trigeminal territory.

Literature

Jacod, M.: "Sur la propagation intracranienne de sarcomes de la trompe d' Eustache, syndrome du carrefour pétrosphenoidal paralysie des 2e, 3e, 5e, and 6e paires craniennes." Rev. Neurol., 28:33-38, 1921.

KEARNS SYNDROME

Synonymy

S. oculocranio-somatic-neuromuscular. S. de Kearns- -Sayre.

Syndromography

Clínica

It begins in childhood before the age of 15. Its clinical picture is characterized by progressive external ophthalmoplegia (ocular muscular dystrophy), pigmentary degeneration of the retina, gaps in the visual field, photophobia and progressive loss of vision; sometimes optic atrophy and cataracts, ataxia, pyramidal signs, decreased or abolished reflexes, alterations in sensitivity, facial and limb asthenia. Conductive hearing loss, otic atrophy.

There are cardiovascular alterations: conduction disorders that cause a tendency to syncope and that generally evolves to complete heart block. Heart failure and often death from cardiac collapse.

The following may occur: scoliosis, dental enamel hypoplasia, hypogonadism, diabetes, hypoparathyroidism, hyperaldosteronism, delayed body growth, gonadal defects and hearing loss.

Paraclinical examinations

Electrocardiogram. Heart block.

Cerebrospinal fluid. Proteins greater than 100 mg/dL, increased pyruvate and lactate levels. Pleocytosis, hyperalbuminorrachia.

Blood. Increase in lactate and pyruvate levels. Hypercholesterolemia, hypomagnesemia.

Muscle biopsy. Abnormal mitochondria in the muscle fiber.

Electrocardiogram. There are frequently conduction disorders, especially complete right bundle branch block and left deviation of the electrical axis. Signs of complete atrioventricular block can be seen.

Syndromogenesis and etiology

Rare and sporadic hereditary myopathy. Its cause is unknown, but there is evidence that familial cases can be transmitted by mitochondrial DNA, not chromosomal DNA.

Ocular muscle dystrophy is hereditary and is transmitted in an autosomal dominant manner.

In relation to the associated manifestations, the etiology is unknown.

Literature

Beremberg, R. A. et al.: "Lumping or splitting?" Ophthalmoplegia Plus' or Kearns-Sayre syndrome?" Ann. Neurol., 1:37, 1977.

Kearns, Th. P. and G. P. Sayre: "Retinitis pigmentosa, external ophthalmoplegia and complete heart block." Arch. Opthal., 60:280-289, 1958.

Niemeyer, G. and Ch. Huber: "Kearns-Syndrom. Progressive externe Opthamomoplegie, Pigmentdegeneration der Retina und kardiale Reizleitunsstörungen." Schweiz. Med. Wschr., 107:1880-1888, 1977.

KLEINE-LEVIN SYNDROME

Syndromnography

Clínic

It mainly affects adolescent males and is characterized by a diencephalic dysrhythmia: alternation of periods of 4-8 days (3 or 4 times a year) of daytime sleep that lasts several hours or an increase in the duration of nighttime sleep, after which the patient wakes up with voracious hunger. Food intake during the period of hypersomnia exceeds 3 times normal (bulimia). This condition is accompanied by social isolation, negativism, slow thinking, incoherence, lack of attention and memory disturbances.

Syndromogenesis and etiology

Some authors postulate a psychogenic pathogenesis, but without any basis so far.

The etiology is unknown.

KUFS SYNDROME

Synonymy

S. of adult familial amaurotic idiocy. S. of neuronal ceroid lipofuscinosis.

Syndromography

Clínic

The symptoms are the same as those of Tay-Sach disease. The syndrome appears between 15 and 25 years of age. The progression is very slow.

Physical examination. Ataxia, muscle rigidity, and paralysis are found. The retina may be normal or present pigmentary degeneration, absence of a cherry-colored spot in the fundus of the eye that is characteristic of the infantile and juvenile forms of this condition.

Paraclinical examinations

Rectal biopsy. Ganglion cells with stored fat in the Meissner plexus morphologically confirm the diagnosis.

Syndromogenesis and etiology

It is due to metabolic disorders that cause an increase in lipids in the brain (gangliosides). There is a peroxidase deficiency in patients with infantile and juvenile forms of ceroid lipofuscinosis.

It is of genetic etiology and is inherited in an autosomal recessive manner.

Literature

Kufs, H.: "Uber eine Spatform der amaurotischen Idiotie und ihre heredofamilian Grundlagen." Z. Neurol. Psychiat., 95:169- -188, 1925.

KUMMELL SYNDROME

Synonymy

S. compression due to vertebral fracture.

Syndromography

Clinic

It appears months after a major or minor direct or indirect trauma to the vertebrae. Formation of kyphosis (hump) associated or not with neurological symptoms, pain, paralysis of the lower limbs, sphincter disorders.

Syndromogenesis and etiology

The injury caused by the fracture causes degenerative changes in the vertebral structure with bone deformation, but without inflammatory signs.

Literature

Kummell, H.: “Ueber traumatische Ezkrankungen der Wirbelsauld." Deut. Med. Wschr., 21:180-181, 1895.

LAMBERT-EATON SYNDROME

Synonymy

S. of pseudomyasthenia. Myasthenic S. associated with bronchial carcinoma. S. of bronchial neoplasia with myasthenic syndrome.

Syndromography

Clinic

This syndrome occurs in middle age and is characterized by weakness, easy fatigue, mainly of the muscles of the pelvic girdle and thigh, generally accompanied by dry mouth, sexual impotence, thigh pain, peripheral paresthesias with a decrease or abolition of tendon reflexes, eyelid ptosis and diplopia.

The syndrome accompanies the clinical picture of pseudomyasthenia in 7% of cases (the most common, 50%, is lung carcinoma). Unlike myasthenia gravis, the extraocular bulbar muscles are not affected and muscle strength improves with repetitive exercise.
Physical examination. Decreased tendon reflexes, difficulty getting up from a chair or couch.

Signs related to bronchial carcinoma. It can be differentiated from myasthenia gravis, because when performed successively, the maneuver of getting up while the patient is sitting becomes easier in this syndrome; the opposite of what happens in myasthenia, which is increasingly difficult until it is impossible for the patient.

Paraclinical examinations

Electromyography. It shows increased pathognomonic action potential amplitude with intense repetitive stimulation.

Electron microscopy of muscle. It shows an increase in folds of the presynaptic membrane at the myoneural junction.

Chest radiology, bronchography and bronchoscopy. They allow the diagnosis to be specified.

Syndromogenesis and etiology

Set of neurological manifestations of systemic neoplasms (50% of patients with this syndrome have pulmonary neoplasia, oat cells, or anaplastic) whose basic physiological defect is a lack of acetylcholine release in the terminal axons of the motor neuron. This alteration is probably due to substances secreted by tumor cells.

The possibility has been raised that it is due to antibodies directed against a part of the nerve endings, which would result in a decrease in the amount of acetylcholine secreted by the motor plate in response to a nervous stimulus, after observing that mice injected with serum IgG from patients develop electrophysiological and electromicroscopic characteristics of the syndrome.

Literature

Anderson, J.H.; H. D. Churchill-Davidson and A.T. Richardson: "Bronchial neoplasm with myasthenia." Lancet, 2: 1291- -1293, 1953.

Kennedy, W. R.: "The myasthenic syndrome associated with small oat cell carcinoma of lung (Eaton-Lambert syndrome)." Neurology, 18: 757-766, 1968.

Lambert, E. H.: L. M. Eaton and E. D. Rooke: "Defect of neuromuscular conduction associated with malignant neoplasm." Amer. J. Physiol., 187:612-613, 1956.

Prior, C. et al.: "Action of Lambert-Eaton myasthenic syndrome. IgG at mouse motor nerve terminals." Ann. Neurol., 17:587, 1985.

Spencer. A. M., M. S. Sumi and R. Ruff: Paraneoplastic syndromes that involve the nervous system." Curr. Probl.Cancer., 8:4, 1983.

Stolinsky, D.C.: "Paraneoplastic syndromes." West. J. Med., 132:189. 1980.

LATIRISM SYNDROME

Synonymy

S. of lupinosis.

Syndromnography

Clinic

This syndrome is more common in men. Sudden onset, preceded by exposure to cold or humidity (relent). Upon waking up and getting up, the patient complains of pain in the lumbar region, weakness of the lower limbs, and slight fever (sometimes). paresthesias. The weakness, which is slight at first, progresses to permanent spastic paralysis, with loss of bladder and rectal control and impotence. Later they appear: loss of sensitivity to pain and temperature, tendon hyperreflexia and Babinski sign.

Syndromogenesis and etiology

The anatomical lesions that cause the clinical picture are: degeneration of the anterior and lateral cords of the dorsal and lumbar segments of the spinal cord. They are related to the ingestion of legumes of the species Lathyrus sativus and other related subspecies as the main food, in situations of hunger, as occurs in Eritrea, India and Algeria. It has been suggested that the toxic factor in the seeds is aminopropiononitrile. Malnutrition also plays an important role.

Literature

Gopalan, C.: "Lathyrism syndrome." Trans. Royal Soc. Trop. Med. Hyg., 44:333-338, 1950.

Minchin, R.: "Primary lateral sclerosis of South India; lathyrism without lathyrus". Br. Med. J., 1:253-255, 1940.

LAURENCE-MOON SYNDROME

Synonymy

S. of brain-retinal degeneration. S. hypogenital adipose with congenital malformations.

Syndromography

Clinic

The most frequent symptoms in this syndrome are: mental retardation, pigmentary retinopathy, hypogenitalism (microphaly, hypospadias and testicular ectopia in the prepubertal stage) and spastic paraplegia.

Paraclinical examinations

Electroretinogram.

Campimetry.

Kidney ultrasound.

Dermatoglyphs.

Karyotype.

Neurological study.

Syndromogenesis and etiology

This syndrome has a genetic etiology and is transmitted by autosomal recessive inheritance. The basic defect is not known.

Studies of the pituitary gland of affected patients have not revealed morphological or immunocytochemical abnormalities that explain the hypogonadism of these patients.

This syndrome is very common in the population of Kuwait.

Literature

Farag. T. I. and A. S. Teeki: "Bardet-Biedl and Laurence-Moon's syndromes in a mixed Arab population." Clin. Genet., 33:78-82, 1988.

Laurence, J.Z. andR. C. Moon: "Four cases of retinitis pigmentosa occurring in the same family, and accompanied by general imperfections of development." Opth. Rev., London, 2:32-41, 1866.

McLouglin, T. G. and D. R. Shanklin: "Pathology of Laurence-Moon." Bact., 93: 65-79, 1967.

LAURENCE-MOON-BIEDL-BARDET SYNDROME

For years this syndrome was incorrectly called Laurence-Moon-Biedl-Bardet syndrome; currently there are two different syndromes: Bardet-Biedl syndrome and Laurence-Moon syndrome.

LESCH-NYHAN SYNDROME

Synonymy

S. of choreoathetosis-self-mutilation and hyperuricemia. S. of juvenile gout with self-mutilations and choreoathetosis.

Syndromography

Clinic

It begins early with bad mood and fits of anger with compulsive self-mutilation mainly of the lips due to modesty, followed later by spasticity choreoathetosis and tremor.

Language learning is late and dysarthric accompanied by moderately severe mental retardation. In patients over 10 years of age, gouty tophi appear in the ears and gouty nephropathy may occur.

Physical examination. Mutilations of lips and fingers, behavioral disorders, choreoathetosis, convulsions, diparesis or quadriparesis with hypertonia and hyperreflexia. Marked mental retardation, intelligence quotient between 30 and 60, hematuria due to urolithiasis that can lead to uremia.

Paraclinical examinations

Blood. Elevated uric acid, 7-10 mg%. Low activity of the enzyme hypoxanthine-guanine-phosphoribosyltransferase (HGFRT). Megaloblastic anemia.

Kidney biopsy. Gouty nephropathy lesions with uric acid deposits in the interstitium are seen.

Syndromogenesis and etiology

There is hyperproduction of uric acid due to "almost complete" hypoxanthine-guanine phosphoribosyltransferase deficiency.

Rare metabolic disorder that is inherited with a recessive trait linked to the XY chromosome and is only suffered by males, while females can be heterozygous carriers.

The gene has been located at Xq26-27. Molecular studies have shown heterogeneity in the syndrome due to point mutations, small DNA deletions that affect the transcription, stability or integrity of the genetic message for the normal synthesis of HGFRT.

Literature

Francke, U.; B. Bakay and W. L. Nyhan: "Detection of heterozygous carriers of the Lesch-Nyhan syndrome by electrophoresis of hair roots lysats." J. Pediat., 82: 472, 1973.

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Keliey, R. et al.: Hypoxanthine-guanine phosphoribosyltransferase deficiency in gout." Ann. Intern. Med., 70:155, 1969.

Sinnet, D. et al.: Lesch-Nyhan's syndrome; molecular investigation of three french canadian families using a hypoxantine-guanine phosphoribosyl transferase cDNA probe." Hum. Genet., 81:4-8, 1988.

Wilson, J.M. et al. : "Hypoxanthine-guanine phosphoribo-syltransferase deficiency. The molecular basis of the clinical syndromes." N. Engl. J. Med., 309:900, 1983.

CERVICAL SPINE INJURY SYNDROME WITH PROGRESSIVE CONFUSION

Syndromography

Clinic

It occurs in patients with fractures of the cervical spine and in non-traumatic spinal cord injuries. There is loss

of memory, hallucinations, delirium and physical signs related to spinal cord injury.

In some patients circulatory disorders appear and then respiratory disorders. There is slight cyanosis.

Paraclinical examinations

Cervical spine x-ray Lumbar puncture.

Syndromogenesis and etiology

It is due to a disruption of motor and sensory pathways, usually as a result of a brain contusion.

It occurs in traumatic fractures of the vertebrae, but can also be observed in hematomyelia, sarcomas and other spinal cord diseases.

Literature

Putnam, T.: "The progressive confusional syndrome following injuries to cervical portion of the spinal cord." Science, 86:542- -543,1937.

METACHROMATIC LEUKODYSTROPHY SYNDROME OF CHILDHOOD

Synonymy

S. de Von-Bogaert-Nijssen.

Syndromography

Clinic

Late infantile form. It occurs in both sexes between the first and fourth years of life. It is characterized by the development of normal locomotion and language, then appearing progressive weakness and hypotonia of the lower limbs, ataxia and spastic paralysis, seizures, optic atrophy (30% of cases), less marked in the juvenile and adult forms. In early stages there is a decrease in muscle tone, absence, decrease or increase in reflexes. In late stages, muscle tone increases, and the Babinski sign is present.

In the final stages, a decerebrate posture occurs, loss of reaction to visual and auditory stimuli.

Youth form. It appears between 4 and 14 years old. The first symptoms are disorders in school performance, emotional and visual.

Adult form. It begins with manifestations of a psychological nature. The patient begins to show loss of interest in work and activities, megalomania, hallucinations, violent reactions and dementia, accompanied by motor disorders that set in months or years after the onset of the disease.

In the late stages of the disease, when the patients are prostrate, genu recurvatum has been observed.

Paraclinical examinations

Blood. Marked decrease or absence of arylsulfatase A, in serum and white blood cells and in cultured fibroblasts.

Peripheral nerve biopsy. The stored material is sulfatides, which are dyed orange-brown and not purple with aniline dyes.

Cerebrospinal fluid. Proteins above 100 mg%.

Syndromogenesis and etiology

The syndrome is caused by an enzyme deficiency, with storage of sphingolipids. The basic disorder is the absence of the enzyme arylsulfatase A, which prevents the conversion of sulfatides into the corresponding cerebroside and therefore conditions the accumulation of sphingolipids.

It is transmitted in an autosomal recessive manner. The human arylsulfatase A gene has been located at 22q13 and has been completely sequenced. This has allowed us to identify several mutations for each clinical type, including the pseudodeficiency mutations of this enzyme.

Literature

Austin, J.; D. Armstrong: yes. Fouch et al.: “Metachromatic leukodystrophy (MLD)." Arch. Neurol.. 18:225-240, 1968.

Greene, H.; G. Hug and W. K. Schubert: “Arylsulfatase A in urine and metachromatic leukodystrophy". J. Pediat., 71:709-711, 1967.

Stein, C. et al.: "Cloning and expression of human arylsulfatase A." J. Biol. Chem., 264:1252-1259, 1989.

LICHTHEIM SYNDROME

Synonymy

S. neuroanemic. S. neurohemaic. S. of funicular myelosis. S. of anemic medullosis. Subacute combined degeneration of the spinal cord. Spinal funicular disease. Pseudotabes anemica.

Syndromography

Clínica

Demyelinating degeneration of the spinal cord, almost always combined (seizure of several spinal cords), which appears linked to Biermer's pernicious anemia and gastric achlorhydria. It generally occurs after the age of 40 and in 70% of patients, associated with pernicious anemia.

The first symptom is paresthesia (tingling in the hands), which is sometimes the only symptom regardless of the degree of anemia. It may be accompanied by disorders of the tactile sensitivity of some fingers. It manifests itself as a burning, urgency or burning sensation in the mouth and tongue with glossitis. Later, asthenia, easy fatigue with light exertion and dizziness appear. At the end of this period, the motor disorder sets in that predominates over the sensory one.

Physical examination. The patient adopts a paretospastic or ataxospastic gait (incoordination) that gives the appearance of a tabetic, which is why it has been called anemic pseudotabes.

The Romberg sign may be absent, doubtful, or positive, but upon pupillary examination the Argill-Robertson sign typical of tabes is not found. As the disease progresses, the neurological condition becomes increasingly worse, paralysis and motor incoordination force the patient to remain in bed. This state is accompanied by neuralgia in the lower limbs, or belt-shaped pains, reminiscent of the shooting pains of tabetic. There are alterations of deep sensitivity, of which the most affected is paleesthesia.

The presence of hypermetry, tremor or other cerebellar symptoms is not uncommon. Sometimes there is spastic or flaccid paraplegia.

The tendon reflexes, mainly patellar and Achilles, are increased and sometimes there is clonus (when lateral cords are taken).

Digestive disorders are rarely absent, mainly achlorhydria. In more advanced periods they appear

sphincter disorders (urinary incontinence): sensitivity disorders (touch, pain and temperature) as well as deep sensitivity are intensified. The psychic sphere may be affected (delirium of persecution).

Paraclinical examinations

Blood. The diagnosis is confirmed by finding a low concentration of vitamin B12 in the serum and a positive Schilling test.

Syndromogenesis and etiology

This syndrome represents a nutritional disturbance of the spinal cord caused by endogenous deficiency, in most cases due to an absorption disorder, of an essential factor for the normal nutrition of the nervous system that is part of the vitamin B complex (B12).

In vitamin B12 deficiency, neurological complications occur, the exact production mechanism of which still constitutes an enigma. Cobalamin participates in the isomerization of methylmalonylcoenzyme A to succinylcoenzyme A, requiring adenosylcobalamin as a prosthetic group of the enzyme methylmalonyl-Co A mutase. Therefore, a B12 deficiency causes high levels of methylmalonate, which is excreted in the urine as methylmalonic acid. Disruption of the succinyl pathway with the progressive accumulation of methylmalonate and propionate (a precursor) can lead to the formation of abnormal fatty acids, which would be incorporated into lipids inside neurons. This biochemical abnormality predisposes to myelin rupture and thus some of the neurological complications that occur in vitamin B12 deficiency would occur.

An alternative explanation is suggested by experiments in which monkeys exposed to nitrous oxide develop a neuropathy clinically similar to vitamin B12 deficiency neuropathy in man. Nitrous oxide inactivates an enzyme necessary for methionine synthesis, so by adding methionine to their diet, exposed animals can protect themselves from developing neural changes. As vitamin B12 is an essential cofactor in the generation of methionine, it is postulated that its deficiency or that of its products may be the common denominator responsible for the neurological alterations in pernicious anemia and in experimental animals exposed to nitrous oxide.

Lichtheim syndrome is almost always (70%) associated with pernicious anemia, but it can be found in vitamin deficiency, pellagra, hemolytic jaundice, sprue, leukemia, alcoholism, diabetes and chronic malaria.

Literature

Dinn, J. J. et al.: "Methylgroup deficiency in nerve tissue: A hypothesis to explain the lession of subacute combined degeneration." J. Med. Sci., 49:1, 1980.

Lechtheim, L: "Zur Kenntniss de perniciosen Anämie." Verhandl. d. Cong. Innere Med., 6:84-96, 1889.

LIST SYNDROME

Synonymy

S. of herniation of the cerebellar tonsils.

Syndromography

Clinic

Severe condition characterized by recurrent headache, vertigo, tinnitus, nausea, progressive vagal bradycardia due to vagal delay of the heart, arterial hypertension (occurs when intracranial pressure begins to approach diastolic blood pressure), causes bulbar ischemia, general compensatory vasoconstriction and finally central respiratory failure.
Cushing considered tonic extension with arching of the neck and back, extension and internal rotation of the limbs with respiratory alterations, cardiac irregularity (bradycardia or tachycardia) and loss of consciousness as a typical sign of cerebellar herniation. The generalized elevation of intracranial pressure, if severe, leads to depression of brain function, resulting in a decrease in psychocerebral activities:
apathy, drowsiness and lack of attention.

Syndromogenesis and etiology

Frontal tumors or general brain edema. It is rarely associated with herniation of the temporal lobe towards the tentorium cerebellum.

There is a downward displacement of the inner and lower part of the cerebellar hemispheres (mainly the ventral paraflocculus or tonsils), through the foramen occipital towards the cervical cord; It can be unilateral or bilateral.

Literature

List, C. .: “Neurologic syndromes accompanying developmental anomalies of occipital bone, atlas, and axis." Arch. Neur. Psych.. 45:577-616, 1941.

PREFRONTAL LOBE SYNDROME

Syndromnography

Clinic

This syndrome begins insidiously and is characterized by personality changes, which are reflected by lack of concern for any situation, social indifference, placidity, lack of aggressiveness, instability and emotional superficiality, sometimes childish excitement occurs (Jastrowitz's moria), jokes and wordplay (Oppenheim's Witzelsucht). Deterioration of intelligence with lack of concentration that prevents executing planned activities with loss of memory. Abulia, decrease in all mental processes and, to an extreme degree, akinetic mutism. Motor disorders that are reflected by difficulty in walking, frontal lobe ataxia or Brun's ataxic gait, abnormal postures, release of grasping and sucking reflexes and incontinence of the sphincters.

Syndromogenesis and etiology

Lesions of the left frontal lobe alter verbal fluency and cause a greater degree of obstinacy, and those of the right side, disorders of learning and visual patterns of space, causing a lack of perseverance. In left frontal lesions, intelligence is reduced more (10 points on the IQ scale) than in the right, perhaps due to a reduction in verbal ability.

It is observed in: frontal leucotomies, meningiomas, tumors of the frontal region, vascular and traumatic injuries, etc.

Literature

Sachs, E.: “Symptomatology of a group of frontal lobe lesions. Brain, 50:474-479, 1927.

Schwab, S. I.: "Changes in personality in tumor of frontal lobe." Brain, 50:480-487, 1927.

MA CKENZIE SYNDROME

Synonymy

Jackson-Mackenzie S. S. of dysphagia-dysphonia.

Syndromography

Clinic

This syndrome is closely related to Jackson's syndrome and is characterized by hoarseness, partial aphonia (unilateral paralysis of a vocal ligament), dysphagia, velopalatine hemiplegia, and partial paralysis of the tongue.

Syndromogenesis and etiology

Lesions of the hypoglossal nerve (pair XII), nucleus ambiguus or branches of the vagus nerve (pair X).

Literature

Mackenzie, S.: Two cases of associated paralysis of the tongue, soft palate and vocal cord on the same side." Trans. Clin. Soc., London, 19:317-319, 1886.

MARCHIAFAVA-BIGNAMI SYNDROME

Synonymy

S. of primary degeneration of the corpus callosum.

Syndromography

Clinic

It generally affects men during middle and late life, in individuals who have ingested large amounts of wine. Variable course (from a few days to several months). It is characterized by a state of agitation and confusion accompanied by visual, auditory and gustatory hallucinations, delirium tremens, memory disorders, negativism and progressive dementia. Language and gait disorders (ataxia), sphincter incontinence, seizures, grasping and sucking movements and hand tremors.

Syndromogenesis and etiology

It has been considered to have a deficiency etiology, but the exact deficiency factor has not been defined, attributing it to the toxic effects of alcohol; however, it has been observed in abstainers so that alcohol cannot be an indispensable factor. Enzyme failure due to vitamin deficiencies and a vasocirculatory defect have been suggested, but these are only speculations.

Literature

Marchiafava, E. and A. Bignami: "Sopra un alterationazione del corpo calloso osservata in soggetti alcoolisti." Riv. Pat. Nerv. Ment., 8:544-549, 1903.

MARIN SYNDROME (AMAT)

Synonymy

Marcus-Gunn S. inverted.

Syndromography

Clínica

Automatic involuntary reflex of eye closure, pressure of the cornea that produces blinking and rapid movement of the jaw on the contralateral side, sometimes the jaw moves slightly forward, with very rapid and minimal movements.

Syndromogenesis and etiology

Unknown. It can be observed a few weeks after hemiplegic attacks and in cases of amyotrophic lateral sclerosis. It is considered an associated movement between the orbicularis oculi and the lateral pterygoid muscle due to a supra-nuclear lesion.

Literature

Marin (Amat), M.: "Sur le syndrome ou phenomene de Marcus Gunn." Ann. Òccul., 156:513-528, 1919.

MAY-WHITE SYNDROME

Synonymy

S. of cerebellar ataxia and deafness. S. of progressive familial myoclonus.

Syndromography

Clinic

It occurs in both sexes, during childhood or adolescence in normal individuals. It begins with myoclonic seizures that spread more and more, triggered by any banal stimulus, and is accompanied by cerebellar ataxia. At first the patient manifests irritability, character alterations, impulsive actions or visual hallucinations that little by little translate into a profound disorder of cognitive functions. Deafness can sometimes be an early sign. At the end of the disease, cachexia forces the patient to stay in bed with rigidity, hypotonia, abolition of tendon reflexes, acrocyanosis and corticospinal signs.

Paraclinical examinations

Electroencephalogram. There are alterations in the waves.

Test for diagnosing deafness.

Syndromogenesis and etiology

It is a syndrome of genetic etiology; families with evidence of autosomal dominant inheritance and families with consanguinity have been reported, which apparently occur due to autosomal recessive inheritance.

The basic defect is unknown.

Literature

May, D. L. and H. H. White: Familial myoclonus, cerebellar ataxia and deafness." Arch. Neurol., 19:331-338, 1968.

MENINGEAL SYNDROME

Syndromography

Clinic

Its onset is generally rapid and a symptomatological triad is described characterized by: headache, vomiting and constipation.

The headache is intense, located preferably frontal or occipital, which is exacerbated by light, noise, movements, coughing or sneezing and which motivates the patient to moan or sometimes exhale the so-called hydrocephalic or meningeal scream and which forces him to remain immobile to relieve the pain, which radiates along the spine (spinal pain) and is produced by inflammation of the leptomeninges at the spinal level.

Vomiting is sudden, abrupt, sometimes without nausea, of a central type, caused by changes in position when moving the head or sitting down.

Constipation is often accentuated as a consequence of abdominal contracture, but has less semiological value.

Other symptoms may include partial or generalized seizures, mainly in children, photophobia (a common symptom), vertigo, nystagmus and sensation of rotation of objects or of the patient himself. Vasomotor type disorders translated by the appearance, after rubbing, of a very bright and persistent red line (Trousseau's meningitic stripe). It has no semiological value.

Fever is common (39-40 °C), the pulse is unstable and irregular. Dissociation of temperature with pulse (bradycardia) is sometimes absent.

Psychological disorders such as changes in character and psychic reactions (hallucinations and delusions, especially in children).

Physical examination. The most characteristic thing is meningeal contracture, which is a very faithful sign and appears early. The patient adopts an antalgic posture, trying to achieve relaxation of the spinal roots to avoid stretching of the periradicular meninges, characterized by shrinkage of the limbs, tilting of the head backwards, adopting in extreme cases lateral decubitus with the head in hyperextension, legs flexed over the pelvis (rifle trigger attitude).

Kernig sign. With the patient lying in the lateral decubitus position, the trunk is flexed until he is seated. When pressure is applied to the knees to obtain the extension of the legs on the plane of the bed, it is found that this is impossible (the legs are semi-flexed), causing pain in the posterior region of the thighs. With the patient lying down, it is impossible to fully extend the leg on the thigh when the thigh is located at a right angle to the pelvis, and the maneuver is painful.

Brudzinski sign. Brudzinski nape sign. By holding the patient's head and trying to flex it by bringing the chin closer to the sternal manubrium, the legs begin the flexion movement. Contralateral Brudzinski reflex. It is caused by flexing the leg on the thigh and the thigh on the pelvis, originating on the opposite side a flexion movement of the contralateral muscle.

Guillain maneuver. Pinching the quadriceps femoris on one side causes a flexion movement of the contralateral muscle.

Lewinson's sign. By ordering the patient to touch the chest with the chin, open the mouth with the aim of achieving this with minimal flexion of the head and spine.

Tripod sign. When sitting on the bed, the patient adopts a stiff, rigid attitude, leaning on both extended upper limbs forming a tripod with the torso. Brudzinski sign of the cheek. By pinching the cheek the patient raises his arms. Sign of toasts. Turning the head to one side raises the contralateral shoulder.

The contracture of the abdominal muscles produces a retraction of the belly, causing the pan sign.

There may be uniocular external strabismus with diplopia, less often eyelid ptosis, unequal pupils, at the beginning myoic and later mydriatic. Sometimes slight hemifacial contracture and lockjaw with teeth grinding are observed.

Optic neuritis is rare.

Paraclinical examinations

Lumbar puncture. It confirms the diagnosis and may present different characteristics depending on the etiology: increased pressure (50-100 cm of water). Clear and transparent appearance like rock water (viral encephalitis) or cloudy and suppurative depending on the number of cells. Greenish color (pneumococcal meningitis) or yellow (mneningococcal meningitis) and sometimes xanthochromic. Less often hemorrhagic (subarachnoid hemorrhage). The cells can reach 300-400 elements or more with a predominance of polymorphonuclear cells, when the germ has a very active phlogogenic power and mononuclear cells (lymphocytes) in viral meningitis. Albumin: increased. Glucosa:

decreases in bacterial meningitis, normal or high in viral meningitis.

The etiological diagnosis is made by culture of the fluid. Colloidal reactions: denote flocculation in the tubes with high liquid dissolutions, which causes curves with

deviation to the right.

Syndromogenesis and etiology

The most common cause of this syndrome is

primary meningoencephalitis, which can be caused by viruses, bacteria (meningococci, pneumococci, staphylococci, gram-negative bacilli and tubercle bacillus), rickettsiae, fungi, spirochetes (leptospirosis and syphilis) and protozoans (Laverán haematozoan), secondary meningoencephalitis (measles, chickenpox, post-vaccination).

Subarachnoid hemorrhage produces a clinical picture very similar to meningeal syndrome.

The presence of an infectious agent, cell, tumor or blood gives rise to an inflammatory reaction in the pia mater and arachnoid mater that extends the subarachnoid and ventricular spaces through the central and lateral foramina of the ventricle. The meningeal reaction is accompanied by hyperproduction of cerebrospinal fluid, which causes increased intracranial pressure, lesions of the underlying cerebral cortex, and irritation of the spinal roots.

Literature

Llanio, R. and colbs.: Clinical propaedeutics and pathophysiology. 5th. ed. t.II. City of Havana, Editorial Pueblo y Educación, 1996. P.836.

HYPERTROPHIC SPINAL MENINGITIS SYNDROME

Synonymy

S. de Charcot-Joffroy. S. of ascending epidural paralysis.

Syndromnography

Clínica

The clinical picture is characterized by severe pain in the neck and dorsal region of the head, shoulders and upper limbs that intensifies with neck movements. There is atrophy and fasciculation of the muscles in these regions that develop over weeks or months.

Physical examination. Tendon hyporeflexia or areflexia of the upper limbs followed by paresis and spasticity of the

legs with loss of sensation below the level of the injury. There are vasomotor and atrophic changes. It may be accompanied by sphincter insufficiency.

Syndromogenesis and etiology

The syndrome does not have a specific cause; in some cases it appears as a complication of syphilis.

Bibliography

Charcot, J. M. and A. Joffroy: "Deux cas dátrophie musculaire progressive avec lésions de la substance grise et des faisceaux antérolatéréaux de la moelle épiniere." Arch. Physiol., 2:354-367, 1869.

PHANTOM LIMB SYNDROME

Syndromography

Clinic

It occurs after a limb amputation. It occurs in 67% of patients. The patient has the sensation that the amputated limb is still present with sensations of position and movement. The limb is perceived in the position it was in at the time of the accident.

Syndromogenesis and etiology

Neuroma of the cut nerve and other lymphatic mechanisms. Phenomenon of peripheral irritation of the stump.

Literature

Brosius, G. R.; M.D. Calvert and T.D.Y. Chin: "Epidemic phlebodynia." Intern Arch. Med., 108:442-447, 1961.

Pearson, J. S.: "Phlebodynia: a new epidemic (?) disease." Circulation, 7:370-372, 1953.

CENTRAL PROTUBERANCIAL MYELINOLYSIS SYNDROME

Synonymy

S. Adams-Victor-Macall.

Syndromography

Clinic

It is a relatively rare syndrome that mainly affects adults of both sexes, although it has been observed in children.

It is characterized by progressive weakness of the mimicry and tongue muscles that causes swallowing and language disorders, tongue paralysis. Sometimes it is accompanied by pseudobulbar phenomena (emotional lability, pathological crying). Quadriparesis that progresses to flaccid quadriplegia without reflexes, Babinski sign. There is frequently an absence of response to painful stimuli and corneal reflexes, urinary incontinence and respiratory paralysis (more rare).

Paraclinical examinations

It is difficult to establish the diagnosis.

Auditory evoked potentials. If performed in the brainstem, they can help demonstrate impaired function of the pontine pathways.

Syndromogenesis and etiology

It is considered an anatomical variety of alcoholic encephalopathy, but it is also observed in non-alcoholic malnourished people.

It has also been observed in patients with neoplasms and kidney diseases, liver failure and various infectious processes. It has been suggested that it is related to the treatment of dehydration and electrolyte imbalance, much more than to alcoholism and malnutrition per se.

Literature

Victor, M. and R. D. Adams: "Effect of alcohol on nervous system." A. Res. Nerv. Ment. Dis. Proc., 32:526-573, 1953.

CLASSIC MIGRAINE SYNDROME

Synonymy

Common migraine S. S. of familial hemicrania.

Syndromography

Clinic

It is a cyclical, paroxysmal clinical condition that presents with a notable familial character characterized by intense unilateral headaches (80% of cases), which can last from a few seconds to several days. It is more common in females and worsens during critical periods of life (puberty, premenstrual, climacteric); It is accompanied by irritability, nausea, vomiting, photophobia, annoying visual flashes, redness and swelling of the eyelids (sometimes),
constipation or diarrhea.

Although the headache most often occurs in the temples, it can occur anywhere on the head, face and neck. Some patients complain of a prodromal condition before the onset of headache, such as facial pallor or other cranial vasomotor disorders such as vertigo.

There are rarely more serious neurological disorders.

Syndromogenesis and etiology

A general vascular abnormality has been confirmed and the extracranial vessels show more anatomical variations than in normal individuals. During the prodromal migraine attack, the intracranial arteries are constricted, and during the headache this contraction is more intense.

For some time now, humoral agents have been sought that could be the basis of this syndrome. It has been observed that the fluid obtained from edematous places in points of headache and maximum hypersensitivity during the crisis, contains a vasodilator polypeptide of the bradykinin type, which lowers the pain threshold and may intervene by promoting sterile local inflammation. However, neither kinins, nor histamine, nor substances such as acetylcholine explain the manifestations of the disorder.

The possibility has been raised that some abnormality in the serotonin metabolism disorder is involved in triggering the syndrome.

This has been based on the observations that the serotonin concentrations that exist before the headache, decrease spontaneously shortly before the onset of the crisis and during this there is an increase in the amount of serotonin metabolites.

serotonin. A platelet disorder has also been raised.
A transient decrease in platelet monoamninoxidase activity, as well as an increase in platelet aggregation, has been observed; however, the significance of these findings is not yet clarified.

Literature

Álvarez, W. C.: "Was there sick headache in 3,000 B.C.?" Gastroenterology, 5:524, 1945.
Friedman, A. P.: "The migraine syndrome." Bull. N.Y. Academic Med., 44:45-62, 1968.

OPHTHALMOPLEGIC HEMIPLEGIC MIGRAINE SYNDROME

Syndromography

Clínic

This syndrome occurs in young adults. It is characterized by moderate hemicrania accompanied by extraocular palsy of cranial nerves II, IV or VI, frequently followed by hemiparesis (3 or 5 days after the onset and when the pain subsides). It may be accompanied by a cranial murmur. The patient recovers within a few days.

Paraclinical examinations

Carotid arteriography.

Syndromogenesis and etiology

The etiology is unknown; in some cases an internal carotid aneurysm has been found.

Literature

Ad hoc Committee: Classification of headache. J.A.M.A. 179:717-718, 1962.

MILLS SYNDROME

Synonymy

S. of the hemiplegic form of amyotrophic lateral sclerosis.

Syndromography

Clínic

It affects the leg and then, in an ascending and gradual manner, it affects the entire limb and also the upper limb on the same side.

Muscle atrophy without fibrillation.

During the course of the disease, extrapyramidal signs may appear.

Syndromogenesis and etiology

Spiller and Mills considered it as a paralysis of cerebral origin due to abiotrophic processes (form of senile paralysis). The characterization of this clinical entity is given by the exclusive capture of the motor neuron. Many different agents show this selective clinical action. Cortical atrophy and ventricular dilation.

Bibliography

Mills, C. K. and W. G. Spiller: "On Landry's paralysis, with the report of a case." J. Nerv. Ment. Dis., 25:365-391, 1898.
Spiller, W. G. and W. T. Llongcope: “Multiple motor neuritis including Landry's paralysis and lead palsy with reports of cases." Med. Record, 70:81-88, 1906.

MYOCLONIC SYNDROME

Syndromography

Clínic

Myoclonus is a clinical sign characterized by a sudden, rapid and brief, disordered and involuntary muscle contraction with a frequency of 7-25/min, with a rhythmic and recurrent character in several muscles and which may be strong enough to displace the affected limb. It can occur spontaneously at rest, or with voluntary movements. Sensory relationships are other of its prominent attributes. The flashing of a light, a loud sound, or a sudden contact with some part of the body can initiate a jolt. Repeated stimuli can cause a series of myoclonic jerks that grow in intensity until producing an attack and appear in any muscle of the body, most frequently in the lower limbs; They are usually characterized by being symmetrical and bilateral, but they can occur on only one side.

Syndromogenesis and etiology

It is a sign present in a wide variety of generalized metabolic and neurological disorders (myoclonus), in the course of encephalitis, Creutzfeldt-Jakob disease, or encephalopathies secondary to respiratory, chronic renal or hepatic failure, or hydroelectrolye imbalance.

Literature

Lance, J. W.: "Myoclonic jerks and falls; an etiology, classification and treatment." Med. J. Aust.. 1:113-119, 1968.

PALATE MYOCLONOS SYNDROME

Synonymy

S. Spencer. S. of velopharyngeal myoclonus. S. of palatal nystagmus.

Syndromography

Clínic

It is characterized by continuous rhythmic contractions of the palate, frequently associated and synchronous with contractions of the pharynx, larynx, tongue, floor of the mouth, neck and diaphragm, with a frequency of 30-180 /min, and persist during sleep and anesthesia, maintaining its rhythm and speed unchanged. Generally, they are bilateral and symmetrical, but sometimes they are unilateral with synchronization of contractions between the different territories. By opening the patient's mouth, the movements of the soft palate and the movements of the pharyngeal constrictor on its posterior wall can be observed.
On laryngoscopic examination, rhythmic adduction and abduction movements of the vocal folds are observed. Sometimes the eyeballs participate in the form of nystagmus, although the jerks occur at two equal times (unlike nystagmus). When the levator muscles of the larynx are taken, their twitches are visible in the neck.

This syndrome is frequently associated with tremor of the the hand

Syndromogenesis and etiology

Tumors, vascular disorders of the brain stem that take the inferior olivary and olivodentate connections.

Literature

Politzer (1862) quoted by J. Gallet: Le nystagmus du voile le syndrome myoclonique de la calotte protuberantielle. Thesis, Paris, 1927.

TUBULAR MYOPATHY SYNDROME

Synonymy

S. of centronuclear myopathy.

Syndromography

Clínica

It affects both sexes. It appears in early childhood or early adolescence, although exceptionally symptoms are observed during the first year of life, but the onset of walking may be delayed. It is characterized by symmetrical paresis of the limbs, a waddle gait (basically the striated muscles are affected), the muscles remain thin and areflexic throughout life. At an older age, the patient presents with a long, narrow face, clubfoot, and scoliosis.

External ophthalmoplegia and eyelid ptosis.

Paraclinical examinations

Electromyography. Abnormal, small motor unit potentials with excessive recruitment, accompanied by fibrillations and acute positive potentials.

Muscle biopsy. Fibers with rows of central nuclei, frequently surrounded by a clear perinuclear zone. Type I fibers are preferentially affected and are atrophic.

Syndromogenesis and etiology

It can be sporadic, but it can be inherited in an autosomal dominant, recessive, or X-linked recessive manner. It appears that muscle development is arrested in the myotubular stage, although the nature of the pathological process is currently not well known.

Literature

Spiro, A. J.;G M. Shy and N. K. Gonatas: "Myotubular myopathy." Arch. Neurol., 14:1-14, 1966.

BIFACIAL MOEBIUS SYNDROME

Synonymy

S. of congenital facial diplegia with convergent strabismus. S. of oculofacial paralysis. S. of congenital paralysis of the VI and VII cranial nerves. S. of nuclear agenesis. S. of oculofacial-bulbar paralysis.

Syndromography

Clinic

It occurs from birth. There is facial paralysis and complete closure of the eyes, inability to abduct the eye further. beyond the midpoint, difficulty in nutrition due to atrophy or deformity of the tongue. Absence of pectoral muscles, syndactyly. Sucking disorders, eversion of the lower lip and open mouth.

Syndromogenesis and etiology

Congenital, possible failure of the development of the cells of the motor nucleus of the facial nerve (cranial nerves VI and VII) and/ or primary defect of the muscles derived from the first and second branchial arches. The term Moebius syndrome should be used only for cases with congenital paralysis of the VI and VII cranial nerves that present skeletal defects. Familial cases have been reported suggesting autosomal dominant inheritance.

Literature

MacDermot, K. D. et al.: "Oculofacial bulbar palsy in mother and son: review of 26 reports of familial transmission within the 'Moebius spectrum of defects." J. Med. Genet., 28:18-26, 1991.

Moebius, P. J.: "Ueber angeborene doppelseitige Abduces-Facialis- - Lahmung." Munchen Med. Wschr.. 31:91-94: 108-111, 1888.

MORVAN SYNDROME

Syndromography

Clínic

It begins insidiously during the second decade or later, characterized by a symmetrical sensory loss, mainly in the lower limbs, of a dissociated type (loss of thermal and pain sensitivities with preservation of tactile and pressure sensitivity).

It is frequently associated with painless infections of the toes, as well as perforating ulcers on the soles and mutilation of the limbs, hyperhidrosis and nail disorders.

Syndromogenesis and etiology

Hereditary neuropathy with an autosomal dominant character. Chronic and selective degeneration of the sensory neuron. Some authors consider it a clinical form of syringomyelia.

MOYAMOYA SYNDROME

Syndromography

Clínic

The condition is mainly seen in infants, children and adolescents (of Japanese origin) of both sexes. It is characterized by weakness of a limb or the arm or leg on the same side. It is accompanied, less frequently, by headaches, seizures, impaired mental development, visual disturbances and nystagmus. Language disorders, sensory disorders, involuntary movements and unsteady gait may occur.

Syndromogenesis and etiology

The term moyamoya (Japanese word for a cloud of smoke or fog) refers to an extensive basilar cerebral rete mirabile (network of small anastomotic vessels at the base of the skull) around and distal to the arterial circle of the brain on the basilar surface. Two possibilities arise: either

that it is a congenital vascular malformation, or collateral vascularization secondary to congenital hypoplasia or acquired stenosis or occlusion of the internal carotid artery in early stages of life that causes abundant small collaterals from the ascending pharyngeal and middle meningeal arteries, belonging to the external carotid system.

Literature

Nishimoto, A. and S. Takeuchi.: Moyamoya disease. Vascular disease of the nervous system. Handbook of clinical neurology. ed. Q. J. Vinken. Amsterdam, North Hollans, 1972.

MUNCHMEYER SYNDROME

Synonymy

S. of myositis ossificans progressiva.

Syndromography

Clínic

It is a rare syndrome that appears in childhood, adolescence or in young individuals. It begins with temporary and localized swelling in the neck and back, later producing progressive apicocaudal ossification of the muscles, fascia and tendons that cause rigidity of movements, causing synostosis and curvature of the trunk. Calcified bridges between adjacent muscles and across joints cause scoliosis, stiffness of the spine, jaw and limbs and limit thoracic expansion. Hard masses with a stony consistency are palpated in the muscles. This rare disorder is spectacular for its consequences; it produces the "stone man" that sometimes appears in circuses. Anomalies such as hypoplasia or malformation of the big toe and thumb and less frequently of other fingers are frequently associated.

Paraclinical examinations

X-ray. Inclusion of dense bone shadows in the paraskeletal muscle masses.

Muscle biopsy. It reveals an intense proliferation of interstitial connective tissue with little cellular inflammatory reaction. Subsequently, the formation of osteoid and cartilaginous tissue occurs, developing in the connective tissue and encompassing relatively intact muscle fibers.

Literature

Basset, A.L.; A. Donath et al.: Diphosphonates in the treatment of myositis ossificans." Lancet, 2:845, 1969.
Lutwat, L.: Myositis ossificans progressive." Am. J. Med., 37:269.1964.

MURRI SYNDROME

Synonymy

S. of generalized cerebellar atrophy. S. of primary cerebellar parenchymal degeneration. S for presenile cerebellar ataxia.

Syndromography

Clinic

It is a rare condition, it predominates in males after the age of 30 (late cerebellar atrophy of the French). It is characterized by ataxia, asynergia and adiadochokinesia accompanied by scanned speech and nystagmus.

Syndromogenesis and etiology

Uncertain. Toxic factors (alcoholism) are suggested.

Literature

Murri, A.: “Degeneration cerebellare da intossicazione endogena.” Riv. Crit. Clin. Med. 1:593 609, 1900.

BODY NECROSIS SYNDROME

FLUTED

Synonymy

S. of symmetrical and bilateral necrosis of the striatum.

Syndromography

Clínica

This syndrome frequently follows a severe epileptic seizure and a state of acute cerebral anoxia. The initial symptoms are: loss of consciousness, absence of involuntary movements, swallowing disorders, athetoid movements.

Syndromogenesis and etiology

Necrosis of the striatum due to anoxic damage, which produces necrosis of the nerve cells of the putamen and the caudate nucleus without destruction of the astroglial architecture or vascular architecture.

Literature

Marinesco, G. and S. Dragenesco: "Contribution anatomo-clinique a létude de syndrome de Foerster." Encephale, 24:685, 1924.

PALLETIGUNAL NECROSIS SYNDROME

Synonymy

S. de Vogt. S. of monoxide anoxia.

Syndromography

Clinic

It occurs in patients who have suffered an episode of coma due to inhalation of carbon monoxide, nitrous oxide or cooking gas. About three weeks later, they recover completely from the coma or slight confusion persists. Relapse can begin weeks after the first episode of coma, and is characterized by an akinetic state, generalized rigidity, extension of the lower limbs, and flexion of the upper limbs. In a minority of cases, there is only slight parkinsonian rigidity.

Syndromogenesis and etiology

Necrosis of the globus pallidus is associated with a lesser or greater degree of cortical damage due to gas toxicity and the intensity of hemorrhagic necrosis.

Literature

Meyer, A.: Ueber der Wirkung der Kohlenoxydvergiftung auf das Zentralnervensystem." Z. Ges. Neurol. Psychiat., 112:172, 1926.

NEOCEREBELLAR SYNDROME

Synonymy

S. of the posterior cerebellar lobe.

Syndromography

Clinic

It appears at any age and in both sexes. It is characterized by ipsilateral or generalized hypotonia, pendulum reflexes, tremor during voluntary movements and static tremor, standing disorders, spontaneous deviation of the lower limbs (tendency to fall towards the side of the injury), asthenia, dysmetria, adiado-chokinesia (the normal rhythm of movements is altered by irregularities in strength and speed), language disorders that can take different forms: syllable-eating dysarthria. "scansion" dysarthria (the patient cuts words into syllables in a similar way to how a verse is scanned in metric), slow, choppy and explosive speech accompanied by writing disorders.

Syndromogenesis and etiology

Vascular or neoplastic, inflammatory or traumatic lesion of the posterior lobe or lateral part of the cerebellar body.

Literature

Holes, G.: "The symptoms of acute cerebellar injuries due to gunshot injuries." Brain, 40:461-535, 1917.

ARNOLD NERVE SYNDROME

Synonymy

S. of Arnold's neuralgia. S. of neuralgia of the auricular branch of the vagus. S. of Arnold's occipital neuralgia.

Syndromography

Clínica

It frequently occurs between the ages of 40 and 70, characterized by pain that begins in the suboccipital region and radiates to the occipital region or to the shoulder, stabbing, paroxysmal, with a tearing sensation. The painful points of the occipital major are located at the midpoint of a line that joins the mastoid process with the atlas. The painful points of the occipital minor are located between the insertions of the trapezius and the sternocleidomastoid at the level of the mastoid process.

Neuralgic crises can be triggered by coughing, the coldness of the pillow or a small head blow.

Syndromogenesis and etiology

This syndrome is produced by local processes of the first two cervical vertebrae: polyradiculitis, consecutive to hypertrophic cervical osteoarthritis, ankylopoietic spondylitis and cervical Pott's disease.

Bibliography

Alayza Escardo, E: “Arnold's neuralgia". JAMA., 161:391, 1956.

ANTERIOR ETHMODAL NERVE SYNDROME

Syndromography

Clinic

Clinical picture characterized by intense bilateral pain in the frontal, parietal, and occipital regions, which does not subside; neck stiffness, which persists for one or more days; areas of tenderness around the ethmoidal foramen and over the nerve.

Syndromogenesis and etiology

It is caused by a hypersensitivity reaction due to intra-tissue pressure in the anterior ethmoidal foramen, a pressure that causes interference with normal nerve conduction.

Literature

Burnham, H. H.: "Anterior ethmoidal nerve syndrome; referred pain and headache from lateral nasal wall." Arch. Otolaryng., 50:640-646, 1949.

ATYPICAL FACIAL NEURALGIA SYNDROME

Syndromography

Clinic

It prevails in young or middle-aged women with long histories of persistent pain attacks with remissions, which is located in the lower half of the head and skull, in the jaw near the posterior molars and which may be accompanied by a sensation of very intense throbbing in the neck, but which are nothing more than headaches of vascular origin.

Syndromogenesis and etiology

It is observed in the course of conversion hysteria and in vascular disorders. Dilation and distension of the extracranial portion of the middle meningeal artery and other branches of the external carotid artery occur.

Literature

Harris, W.: Neuritis and neuralgia. Oxford, London, 1926.

"An analysis of 1 433 cases of paroxysmal trigeminal neuralgia (trigeminal-tic) and end results of grasserian alcohol injection." Brain, 63: 209-224. 1940.

MULTIPLE SENSORY NEURITIS SYNDROME

Synonymy

S. of multiple cutaneous neuritis.

Syndromography

Clinic

Minimal and practically asymptomatic for a long time. Changes in skin sensitivity from hypoesthesia to anesthesia. Spontaneous pain rarely stimulated by slight trauma to the skin, the distribution of the affected areas being disseminated but never symmetrical, more frequently involving sensitive nerves of the fingers and limbs such as the saphenous, lateral femoral cutaneous, and lateral calcaneal nerves. The motor nerves are never affected. Absence of vasomotor or trophic manifestations. There are no systemic disorders.

Syndromogenesis and etiology

It can be an expression of multiple sclerosis or many other diseases.

Literature

Schlesinger, H.: Ueber Neuritis multiplex cutaneous." Neurol. Centralbl.. 30:1218-1221, 1911.

Wartlenberg, R.: "Multiple sensory neuritis: a clinical entity." Trans. Amer. Neurol. Ass., 71:101-104, 1946.

FAMILIAR NEUROBLASTOMA SYNDROME

Synonymy

S. of congenital neuroblastoma.

Syndromography

Clinic

It usually begins before the age of 5 (80% of cases), some later. Usually, its first manifestation is an abdominal tumor mass located in the adrenal or neighboring sympathetic ganglia, which frequently crosses the midline. The clinical picture varies according to its location. At the level of the cervical region it appears in the form of a hard, lobular mass, which affects the posterior triangle of the neck or extends to the sternocleidomastoid region. Chest pain and cough when located in the mediastinum. Urinary or rectal obstruction if the location is in the pelvis. Generally, the first symptoms are more attributable to metastases than to the primary neoplasia.

Paraclinical examinations

Urine. Elevated levels of catecholamines of one or more of its derivatives 3-methoxy-4-hydroxymandelic acid (VMA) and hemovalinic acid (HVA).

Radiology. It allows the localization of the tumor and bone metastases that are frequently bilateral, symmetrical with areas of destruction and proliferation of new bone.

Computerized axial tomography. It shows a hypodense mass whose density increases uniformly after the administration of contrast substance, with a variable degree of hemorrhage and calcification.

Cerebrospinal fluid. Cytology.

Syndromogenesis and etiology

The possibility that it is transmitted by autosomal dominant inheritance is suggested. It is one of the most common neoplasms in children. More than half originate in the adrenal gland or in the sympathetic ganglia, and can appear at any point in the sympathetic ganglia chain or can come from the spinal ganglia. On rare occasions it occurs as a primary tumor of the central nervous system.

In human neuroblastoma cell lines, it has been possible to isolate genomic DNA homologous to the MYC oncogene, but amplified; the MYCN oncogene that has been located at 2p24. There are two chromosomal alterations related to this oncogene: a long chromosomal region of homogeneous color and the presence of double ninuts that never coexist in a cell.

Literature

Brodeur, G. M. and R. C. Seager: "Gene amplification in human neuroblastomas: basic mechanism and clinical implications. Cancer Genet. Cytogenet., 19:101-111, 1986.

Chatten, J. and M. L. Voorhess: "Familial neuroblastoma." N. Engl. J. Med. 277:1230-1236, 1967.

Dodge, H. J. and M. C. Benner: “Neuroblastoma of adrenal medullary in siblings. " Rocky Mount. Med. J., 42:35-38, 1945.

HEREDITARY AMYLOIDOSIS NEUROPATHY SYNDROME TYPE I

Synonymy

Andrade type hereditary amyloidosic neuropathy. S. Portuguese type hereditary amyloidosis neuropathy.

Foot disease. S. of amyloidosic neuropathy type I.

Syndromography

Clínic

It affects both sexes equally and begins between the third, fourth and fifth decades of life. Orthostatic hypotension precedes other symptoms. There is insidious progression of symmetrical peripheral paresthesias of the lower limbs, which progress slowly and eventually extend to the upper limbs, accompanied by vegetative alterations such as: loss of the pupillary light reflex and miosis, anhidrosis, vasomotor paralysis, diarrhea alternating with constipation and impotence.
Gait disorders (due to alterations in position sense and slight muscle weakness). Abolition of tendon reflexes. Heart rhythm disorders (branch block or atrioventricular block with cardiomegaly). Weight loss, anorexia and hepatomegaly. More rarely it may be accompanied by vitreous opacity, which progresses to blindness and hearing loss.

Paraclinical examinations

Sural nerve biopsy. Decrease in non-myelinated and short myelinated fibers with nodular amyloid deposits along the nerve trunks.

Biopsy of skin, muscle, gums, rectal mucosa or kidney. Amyloid is demonstrated by appropriate stains and electron microscopy.

Syndromogenesis and etiology

It is inherited as an autosomal dominant trait. It mainly affects the short sensory and autonomic fibers. The lumbosacral dermatomes show loss of syringomyelic nociception and thermal discrimination with preservation of touch and pressure.

The defect has been located in the transthyretinolin (TTR) protein gene. This is a transport protein for thyroxine and retinol (vitamin A). The mutation consists of an exchange of valine for methionine at position 30 (val 30 met). The gene has been assigned to chromosome 18.

Literature

Mahloudji, M.; R.D. Teasdall; J. J. Adamkiewicz et al.: "The genetic amyloidosis; with particular reference to hereditary neuropathic amyloidosis, type II (Indiana or Rukavina type)." Medicine, 48:1-37, 1969.

HEREDITARY AMYLODOSE NEUROPATHY SYNDROME TYPE II

Synonymy

Rukovina type hereditary amyloidosic neuropathy. S. Indiana type hereditary amyloidosic neuropathy.

Syndromography

Clínic

It affects both sexes equally during the third to fourth decades of life. It is characterized by symptoms of peripheral neuropathy, which most severely affects the upper limbs, starting in the hand. It is characteristic to observe numbness of the thumb and the first three fingers due to compression of the median nerve, at the level of the carpal tunnel ligament.

Small motor disorders may be observed.

Paraclinical examinations

Sural nerve biopsy. Decrease in short myelinated and unmyelinated fibers with nodular amyloid deposits along the nerve trunks.

Biopsy of skin, muscle, gum or rectum. The amyloid substance is demonstrated by appropriate stains and electron microscopy.

Syndromogenesis and etiology

Familial polyneuropathy of autosomal dominant inheritance whose mutation is at the level of the transthyretinolin protein gene and produces a change from isoleucine to serine at position 84 (see amyloidosic neuropathy type I). Nonhereditary amyloidosis neuropathy syndrome

Syndromography

Clinic

It predominates in men (84%). The age of onset is around the fourth to sixth decades of life. It is characterized by symptoms similar to hereditary amyloidosic neuropathy, although it affects the distal portions of the lower limbs more than the upper limbs. It is accompanied by cardiomegaly (21%), hepatomegaly (16%), splenomegaly (5%), macroglossia and nephropathy.

Syndromogenesis and etiology

Unknown.

Literature

Ostertag, B.: "Familiare Amyloid-Erkrankung." Z. Menschl. Vereb. Konstit-Lehre. 30:105-115, 1950.

NIELSEN II SYNDROME

Synonymy

S. of the cingulate gyrus.

Syndromography

Clinic

There is apathy, akinesia, mutism, incontinence.

Physical examination. Eyes open, normal muscle tone, indifference to pain, bilateral Babinski sign.

Syndromogenesis and etiology

There is bilateral damage to the cingulate gyrus.

The etiology is unknown.

Literature

Ford, F. R.: Diseases of nervous system. ed. 4, Springfield, Thomas, 1960.

Nielsen, J. M.: Agnosia, apraxia and aphasia, Hoeber, New York, 1946.

ANTERIOR SPINAL ARTERY OCCLUSION SYNDROME

Synonymy

S. de Beck. S. of hemianesthetic hemiplegia. S. hypoglossal.

Syndromography

Clinic

It usually presents abruptly, aplopectiform with pain and paresthesia and varies according to the site of occlusion:

1. Occlusion of the anterior spinal artery at the level of the medulla oblongata (Davison syndrome): flaccid quadriplegia, loss of discriminative sensitivity below the level of the lesion.
2. Occlusion of the branches that supply the paramedial area of the medulla oblongata: ipsilateral paralysis of the tongue and contralateral hemiplegia with tactile sensitivity disorder.
3. Occlusion at the thoracolumbar level: segmental muscle atrophy and spastic paralysis of the lower limbs, occasionally unilateral with contralateral hemianesthesia, sensory dissociation below the lesion.
4. Occlusion at the level of the lower portion of the cord: flaccid paralysis of the lower limbs with dissociative changes in sensitivity. Abolition of reflexes of the affected area, disorder of surface sensitivity.

Preserved paleesthesia and bathyesthesia.

Syndromogenesis and etiology

Thrombosis (most common) and neoplasms. Atherosclerosis, syphilis, trauma, coarctation of the aorta.

Literature

Spiller, W.G.: "Thrombosis of the anterior cervical median spinal artery; syphilitic acute anterior polymyelitis." J. Nerv. Ment. Dis., 36:601-613, 1909.

PAGE SYNDROME

Synonymy

S. of cerebral hypertension.

Syndromography

Clinic

It appears in young or middle-aged women, occasionally in men. It is characterized by the periodic appearance of red spots covered by small droplets of perspiration on the face, upper chest, and sometimes on the abdomen. The members during the crisis are cold, pale. There is tearing without emotional cause. Headache, tachycardia and abdominal hyperperistalsis. Emotional polyuria. Blood pressure lability, which increases during attacks. Occasionally low-grade fever.

Syndromogenesis and etiology

Unknown. It is suggested that it is caused by a hypothalamic disorder. A similar syndrome is observed in some cases with tumor compressing the hypothalamus.

Literature

Page, I. H.: “A syndrome simulating diencephalic stimulation occurring in patients with essential hypertension." Amer. J.Med. 190:9-14, 1935.

Schroeder, H. A. and M. L. Goldman: Test for the presence of the hypertensive diencephalic syndrome using histamine. Amer. J. Med. 6:162-167, 1949.

PAINE-EFRON SYNDROME

Synonymy

S. of ataxia telangectasia II.

Syndromography

Clínic

It appears in late adolescence or early youth. It begins with pain in the dorsal region and thigh, later it is accompanied by slowly progressive ataxia. Diffuse telangiectasias and darkening of the nevi.

Syndromogenesis and etiology

Dominant inheritance patterns. Possible spinocerebellar degeneration.

The etiology is unknown

Literature

Paine, R. S. and M. I. Efron: "Atypical variants of ataxiatelangectasia syndrome." Develop. Med. Child. Neurol., 5:14-23, 1963.

WACHSMUTH-LEYDEN PROGRESSIVE BULBAR PALSY SYNDROME

Synonymy

Duchenne S. S. of labioglossopharyngeal paralysis. S. of progressive spastic bulbar palsy.

Syndromography

Clínic

It appears between 50-60 years of age. It begins with progressive language disorders (difficulty pronouncing lingual (r, n, l) , labial (b, m, p, f), dental (d, t) and palatal (k, g) consonants; chewing disorders due to weakness of the chewing muscles (bulldog reflex); swallowing disorders with occasional nasal regurgitation, drooling, loss of pharyngeal and laryngeal reflexes, nasal voice due to difficulty in raising the soft palate, fasciculations of the chin and tongue, which is small and atrophic with impossibility of propulsion due to disorders of the genioglossus muscle. The bipedal position and movements are affected by spasticity of the limbs. The shoulders are raised with difficulty and it is not possible to raise them.

Loss of emotional control with sudden episodes of laughing and crying. Weakness of facial muscles.

Paraclinical examinations

Electromyography. Denervation fibrillations and fasciculations. Reduction in the conduction speed of motor nerves.

Syndromogenesis and etiology

Clinical variant of amyotrophic lateral sclerosis. Degeneration of the nuclei of cranial nerves V, VII, IX,

Literature

Duchenne, G.: "Paralysie musculaire progressive de la langue, duvoile du palais et des levres: affection non encore décrite comme espeche morbide distincte." Arch. Gén. de Méd., 16:283-296, 1860.

SATURDAY NIGHT PARALYSIS SYNDROME

Synonymy

S. alcoholic neuropathic.

Syndromography

Clinic

This paralysis occurs in heavy alcohol drinkers. After alcohol poisoning, the patient wakes up feeling numbness in the upper limb, when the arm is abducted over the edge of a chair or with the head on it. The most frequent paralyzes are those of the muscles innervated by the radial nerve (they prevent the extension of the elbow, wrist and fingers); The peripheral nerves of the lower limbs are also affected, especially the peroneal nerve.

Syndromogenesis and etiology

This syndrome is due to compressive neuritis. Alcoholic neuritis frequently exists previously.

Literature

Grinker, R. R. and A. L. Sahs: Neurology, ed. 6, Springfield, Thomas, 1966.

RAEDER PARATRIGEMINAL PARALYSIS SYNDROME

Synonymy

Trigeminal paralysis in oculopupillary sympathetic syndrome.

Syndromography

Clinic

The symptoms are unilateral and characterized by intense pain concentrated around the eye, which may be continuous with periods of exacerbation and relaxation. Sometimes it may be accompanied by symptoms related to seizure of the parasellar cranial nerves (II, II, IV and VI).

Physical examination. Eyelid ptosis, miosis, and absence of facial sweating are observed (which establishes the difference with Horner's syndrome). Conjunctival injection and tearing are generally present.

Paraclinical examinations

Skull and cervical spine radiology.

Arteriography.

Syndromogenesis and etiology

This syndrome can be produced by neoplastic, traumatic, inflammatory, vascular, or idiopathic lesions that damage the oculosympathetic fibers distal to the bifurcation of the common carotid artery (at this level sweat fibers are scarce).

When there is seizure of the parasellar cranial nerves (Raeder syndrome type I), it occurs due to tumors, abscesses or aneurysms of the anterior part of the middle cranial fossa.

When there is no cranial nerve intake (Raeder II syndrome), it is caused by dental abscesses, aneurysm of the internal carotid artery, etc.

Literature

Law, W. R. and E. R. Nelson: Internal carotid aneurysm as a cause of Raeder's paratrigeminal syndrome." Neurology, 18:43-46, 1968.

Raeder, J. G.: "Paratrigeminal paralysis of oculopupillary symptoms." Brain, 47:149-158, 1924.

THYROTOXIC PERIODIC PARALYSIS SYNDROME

Synonymy

S. of Kitamura.

Syndromography

Clínic

This syndrome is more common in men between the third and fourth decades of life, mainly in individuals of Japanese origin (2.8% of Japanese patients with hyperthyroidism suffer from this syndrome). Appear, general-

mind, early in the morning when you wake up, or during the day after resting after exercise or a large meal. There are painless episodes of muscle paralysis of the trunk and limbs that make the patient unable to get up or move. Eye movements, salivation, without respiratory disorders that last 12-24 hours. Patients present all the symptoms and signs of hyperthyroidism (Basedow syndrome), although these may appear months after the muscular syndrome. In some cases the thyroid gland may be enlarged without other signs of hyperthyroidism (apathetic hyperthyroidism). During crises, the muscle reflexes of the affected muscles are decreased or absent, without an increase in muscle tone. This myopathy disappears when the patient becomes euthyroid with treatment. Paralysis attacks can be induced by 10 units of insulin.

Paraclinical examinations

Electromyography. During the crisis, type myopathy is revealed. paresis.

Glucose tolerance test. Altered.

Hyperthyroidism test. Altered.

Muscle biopsy. Generally, normal. Few fibers undergo degenerative changes.

Optical microscopy. It reveals central subsarcolemmal vacuolization in a small group of the affected muscles.

Syndromogenesis and etiology

Thyrotoxic periodic paralysis is associated with hyperthyroidism. It is assumed that hyperthyroidism externalizes latent forms of Cavaré-Westphal disease (hypokalemic familial periodic paralysis).

Literature

Kitamura, R. and I. Nihonnaukagakukai-Zasshi, quoted by T.

Shinosaki: "Klinische Studien uber die periodischen paralysise derextremitaten." Zkschr. ges. Neurol. Psychiat., 100-564-611, 1926.

Norris, F H., B. J. Panner and J. M. Stormont: Thyrotoxic periodic paralysis; metabolic and ultrastural studies." Arch. Neurol. 19:88-98, 1968.

TODD POSEPILEPTIC PARALYSIS SYNDROME

Syndromography

Clínic

Transient paralysis following a focal motor seizure, as a sequel to the Jacksonian attack, manifested by temporary paresis or paralysis of the limbs lasting a few hours or days.

Syndromogenesis and etiology

Apparently it is produced by exhaustion of the neurons of the epileptic focus.

Literature

Jasper, H. H.: Basic mechanisms of the epilepsies. Little Brown, Boston, 1969.

Todd, R. B.: clinical lectures on paralysis. ed. 2, Churchill, London, 1856.

PSEUDOBULBAR PARALYSIS SYNDROME

Syndromography

Clinic

This syndrome appears after the age of 40, and is characterized by dysarthria, slow, stammering speech and even anarthria, temporary complete aphonia or monotone monotonous voice, dysphagia (choking), difficulty walking with loss of strength in all limbs, reminiscent of a low-intensity spastic tetraparesis, spontaneous laughter and crying that cannot be controlled. Exaggeration of automatic movements and reflexes, facial expression may be similar to that of parkinsonism (mask facies), nasal voice without intonation, low murmur volume, and uncontrollable salivation.

Syndromogenesis and etiology

Atherosclerosis with multiple areas of softening.

Literature

Magnus, A.: “Fall von Aufhebung des Willenseinflusses auf einige Hirnnerven." Arch. Anat. Physiol. or. Wissench. Med., 258- -267. 1837.

PROGRESSIVE SUPRANUCLEAR PARALYSIS SYNDROME

Synonymy

S. de Steele-Richardson-Olszewsky.

Syndromography

Clínica

It is observed more frequently in males during the sixth decade of life (45-73 years). It begins with vague personality changes combined with balance and gait disorders, sudden falls, visual and ocular alterations, constant pseudophthalmoplegia that mainly affects vertical gaze, especially downward, loss of Bell's phenomenon (reflex superior movement of the eyes when the eyelids are forced to close) and ocular convergence and the pupils then decrease in size. In the course of its evolution, horizontal eye movements are also affected, preserving the oculovestibular reflexes, until the total loss of voluntary eye movements.

Disturbance of vertical movements and loss of the rapid component of optokinetic (heat-induced) nystagmus. Dysarthria (slurred speech). Dysphagia. Dystonic rigidity of the neck and upper part of the trunk, the limbs become inflexible with Babinski's sign in some cases, expressionless facies, clumsy gait with slight ataxia of the limbs, decreased memory, retardation of thought processes, apathy and disturbance of the ability to manipulate the available information.

The patient becomes anarthritic, immobile and almost useless with some degree of dementia.

Syndromogenesis and etiology

Loss of neurons and gliosis occurs in the mesencephalic tectum and tegmentum, subthalamic nucleus of Luys, vestibular and ocular nuclei.

The etiology is unknown so far. The possibility of a slow virus is raised, but attempts to transfer it to monkeys through intracerebral inoculation of brain tissue have failed.

It is observed in parkinsonism, pseudobulbar palsy and dementia.

Literature

Chavany, J. A.: L. von Bogaert et S. Godlewski: “Sur un syndrome de rigidité a prédominance axiale avec perturbation des automatismes oculo-palpébraux d'origine encéphalitique.” Presse Med., 59:958-962, 1951.

Di Mauro, S.; E. Bonilla et al.: "Mitochondrial myopathies." Ann. Neurol.., 17:521, 1985.

Maher, E. R. and A. J. Lees: "The clinical features and natural history of the Steele-Richardson-Olszewski's syndrome. Neurology, 36:1005, 1986.

Steele, J.C et al.: “Progressive supranuclear palsy.” Arch. Neurol., 10:333-359, 1964.

PAINFUL PARAPLEXY SYNDROME

Syndromography

Clínica

Extremely severe pain at a specific point in the spine, due to compression of the vertebral root, followed by paraplegia and cachexia.

Paraclinical examinations Spine

Bone survey.

Syndromogenesis and etiology

Osteogenic or osteolytic vertebral lesion, multiple myeloma, prostate, lung, breast, kidney or colon metastasis.

Literature

Grinker, R. R. and A. L. Sahs: Neurology. ed. 6, Springfield, Thomas, 1966.

PARINAUDII SYNDROME

Synonymy

S. of the mesencephalic colliculi (quadrigeminal tubercles). Posterior mesencephalic S. S. of conjugated gaze paralysis. S. of Koerberg-Salus-Elshing.

Syndromography

Clínic

This syndrome is characterized by paralysis of gaze in the vertical plane, the eyeballs cannot be raised or lowered; More frequently, upward movement is affected, while downward movement is preserved. The paralysis is usually conjugate and is usually not accompanied by diplopia but is accompanied by convergence paralysis or paresis and frequent abolition of the photomotor reflex with preservation of accommodation (Argill-Robertson sign). Eyelid retraction.

In the clinical form described by Koerberg-Salus, ocular and eyelid nystagmus is associated with different eye movements.

Syndromogenesis and etiology

This syndrome is caused by mesencephalic lesions that affect the region adjacent to the gray matter, which surrounds the aqueduct of the brain on both sides of the midline and in the region of the posterior cerebral commissure and mesencephalic roof.

The causes are: vascular and tumor lesions (the most common is pinealoma) and multiple sclerosis that affect the oculomotor function.

Literature

Borde, R. M. and P. J. Savino: Clinical Decisions in Neuro- - Ophthalmology, St. Louis, Mosby, 1985.

Hiller, N. R.: Walsh and Hoyt's clinical in Neuro-Ophthalmology. vol. 2 Baltimore, Williams and Wilkins, 1985.

Leigh, R. J. and D. S. Zee: The neurology of eye movement. Davis, Philadelphia, 1983.

Pavan-Langston, D. (ed.): Manual of ocular diagnosis and therapy. ed. 2 Little Brown, Boston, 1985.

ENCEPHALYTIC PARKINSONISM SYNDROME

Syndromography

Clinic

This syndrome appears at any age gradually and has rapid development. It is characterized by slight prodromes that may sometimes be missing and are caused by: lockjaw

moderate pain and headache followed by a state of intense asthenia that leads the patient to complete immobility. The face acquires an expression of fixed gaze with the open palpebral slit that resembles stupor. There are very characteristic seborrheic reflexes that cover the skin with oil, almost constant salivation and frequently unilateral pupillary reaction. The patient adopts a hunched attitude, with the head flexed and the dorsal kyphosis is accentuated, with loss of automatic swing of the upper limbs during walking and is accompanied by anteropulsion. There is dissociation of the components of sleep:
During the day he presents immobility of sleep but without sleeping, while during the night he sleeps, but with marked motor restlessness (gesticulates, speaks, stands up, etc.).

The tremor is violent, almost myoclonic.

Paraclinical examinations

Cerebrospinal fluid. Subtle increase in blood pressure, increase in albumin, moderate lymphocytic pleocytosis.
Pandy's reaction is almost always positive. Negative Wassermann reaction. Hyperglycorrhachia. Luetic-type colloidal reactions with medium intensity flocculation.

Syndromogenesis and etiology

This syndrome is caused by a complication of Von Economo type encephalitis.

It is caused by injury to the substantia nigra of the midbrain and globus pallidus of the telencephalon.

Literature

Grinker, R. R. and A. L. Sahs: Neurology. ed. 6, Springfield, Thomas, 1966.

SUPERIOR CEREBELLAR PEDUNCLE SYNDROME

Synonymy

S. of the anterior superior cerebellar artery.

Syndromography

Clínica

It appears suddenly, without loss of consciousness. Unilateral. It is characterized by involuntary resting movements and cerebellar hemisyndrome that is revealed by lateropulsion during walking, muscle hypotonia, dysmetria and tremor, all of which are ipsilateral in location. Contralateral loss of thermal and pain sensitivity including the head. Sometimes there is contralateral hearing loss and Horner syndrome is present. Normal swallowing persists, there is no paralysis or anesthesia in the area of innervation of the glossopharyngeal nerve, no hoarseness or signs of seizure of the pyramidal tract. The prognosis depends on the extent and nature of the injury. Frequently the evolution is favorable.

Paraclinical examinations

Lumbar puncture.

Computerized axial tomography.

Arteriography.

Syndromogenesis and etiology

This syndrome is caused by obstruction of the anterior superior cerebellar artery due to thrombosis and less frequently due to embolisms, neoplasms and inflammatory processes.

This obstruction causes softening lesions that affect the superior surface of the cerebellar hemisphere on that side, accompanied or not by injury to the superior cerebellar peduncle and the lateral lemniscus.

Literature

Dow, R. S. and G. Muruzzi: The physiology and pathology of the cerebellum. University of Minnesota Press, Minneapolis, 1958.
Porot, A.: “Hémoragie limitée du pédoncle cérébelleux superiéur droit; hémisyndrome cérébelleux diract." Lyon Méd. 106:1137- -1141, 1906.

PELIZAEUS-MERZBACHER SYNDROME

Synonymy

S. of congenital extracortical aplasia. S. of sudanophilic leukodystrophy. S. of Spielmeyer leukodystrophy.

Syndromography

Clinic

It appears exclusively in the male sex. It begins in the first months of life or during childhood. It has a progressive and slow course and is characterized by abnormal, arrhythmic eye movements (pendular, rapid, irregular, frequently asymmetrical nystagmus), intermittent head movements, ataxia, intentional tremor, choreiform and athetotic movements of the arms. Disorders of psychomotor development and mental disorders.

Syndromogenesis and etiology

It is due to defective myelination of the brain, brainstem, cerebellum, spinal cord and peripheral nerves.

It is a syndrome of genetic etiology that is transmitted by autosomal recessive inheritance linked to the X chromosome. The mutation is found in the gene that encodes proteolipid protein (PLP) in Xq22. The absence or defect of this protein produces demyelination and the absence of oligodendrocytes has been observed in the brain of patients with this syndrome. At least six different types of PLP gene mutations have been described.

This syndrome is transmitted by X-linked recessive inheritance, and the biochemical mechanism is unknown.

Literature

Pelizaeus, F.: "Uber eine eigen thumliche Form spastischer Lahmung mit Cerebraler Scheinungen aufghereditarer Grundlage (Multiple Sklerose)." Arch. Psychiat. Nervenkr., 16:698-710, 1885.

Raskind, W.H. et al.: "Complete deletion of the proteolip protein gene (PLP) in a family with linked Pelizaeus-Merzbacher disease." Am. J. Hum. Genet., 49:1355-1360, 1991.
Seitelberger, F.: Pelizaeus Merzbacher disease. In Vinken, P. and G. Bruin, Handbook of clinical neurology. vol. 10 Amsterdam, NorthHolland 1970, pp: 180.
Zeman, W.; W. Demyer and H. F. Falls: "Pelizaeus-Merzbacher disease. A study in nosology." J. Neuropath. Exp. Neurol., 23:334- -354, 1964.

PENFIELD SYNDROME

Synonymy

S. of central brain epilepsy. S. of anterior diencephalic autonomic epilepsy.

Syndromography

Clinic

Syndrome characterized by seizures accompanied by vegetative manifestations: facial congestion, salivation, perspiration, lacrimation, exophthalmia, pupillary dilation, tachycardia,

The seizures are of variable duration (1-15 min), and end with a short period of obtundation.

Syndromogenesis and etiology

It occurs due to a localized injury to the floor and walls of the third ventricle.

Literature

Penfield, W.: “Diencephalic autonomic epilepsy.” Arch. Neurol. Psychiat., 22:358-374, 1929.

POSHERPETIC SYNDROME

Synonymy

S. of postherpetic neuralgia.

Syndromography

Clinic

It generally appears in the elderly, after a zoster infection. It is extremely rare and is characterized by a constant burning sensation, with superimposed waves of intense and prolonged stabbing pain and exquisite sensitivity to the slightest stimulus on the skin of the affected territory.

Syndromogenesis and etiology

Degenerative changes and incomplete regeneration of the nerves.

Literature

Brain, Lord: Diseases of the nervous system. ed. 6. London Oxford University Press, 1962.

DIABETIC PSEUDOTABES SYNDROME

Synonymy

S. of diabetic neuropathy.

Syndromography

Clínic

It is characterized by tendon areflexia, deep anesthesia and pain in the branches of the peroneal nerve (external popliteal sciatic) with muscle atrophy. In some cases, mononeuritis of the branches of the brachial plexus.

Paraclinical examinations

Corresponding to the underlying etiology.

Syndromogenesis and etiology

This syndrome occurs as a complication that appears in the course of diabetes and is due to changes in the nerve metabolism. It mainly affects diabetics aged 50-60 years.

Literature

Jackson, J. H.: "Contribution to the comparative study of seizures." Brain, 9:1-23, 1886.

BRAIN RADIATION SYNDROME

Synonymy

S. of brain necrosis due to radiation.

Syndromography

Clínic

It occurs in patients treated with high doses or multiple courses of ionizing radiation. The syndrome develops 7-12 months after the last treatment and presents variable symptoms: headaches, seizures, paresis, memory impairment, emotional lability, aphasia and stupor.

Syndromogenesis and etiology

It is a degenerative process of nerve cells due to the direct effect of radiation and at least in part to ischemia caused by post-radiation vasculitis.

Autoimmune mechanisms have been thought to play some role in tissue injury.

Literature

Fisher, A.W. and H. Holfelder: Lakales Amyloid im Gehirn Eine Spatfalge von Rontgenbestrahlungen." Deutsch. Z. Chir., 227:475-483, 1930.

RAYMOND-CESTAN SYNDROME

Synonymy

S. of Raymond. S. of Cestan. S. of lateral gaze dissociation. S. alternate superior pons.

Syndromography

Clínic

It is characterized by monolateral paralysis of lateral gaze movements due to seizure of the VI nerve (abduccens or abducens nerve) on the same side, contralateral hemiplegia, hemianesthesia of the face, trunk and limbs. The prognosis depends on its etiology.

Syndromogenesis and etiology

This syndrome is caused by an injury to the upper part of the pons that affects the pyramidal tract, posterior longitudinal band, medial lemniscus, central trigeminal tract after its crossing, olivorubrical and rubrospinal tracts.

Literature

Raymond, F. et R. Cestan: "Troi observation de paralysie des mouvements associés des globes oculaires." Rev. Neurol., 9:70- -77, 1901.

REICHERT SYNDROME

Synonymy

S. of neuralgia of the tympanic plexus. S. of the neuralgia of the geniculate ganglion. S. of Jacobson's nerve.

Syndromography

Clínic

Paroxysm of stabbing pain in the external acoustic meatus, associated with other pains in the face and posteroauricular area.

After section of this nerve, there is unilateral loss of sensations in the soft palate, pharyngeal wall and Eustachian tube, epiglottis and the posterior third of the tongue.

Syndromogenesis and etiology

The etiology is due to inflammatory or neoplastic irritation of the tympanic branch of the glossopharyngeal nerve (Jacobson's nerve).

Literature

Reichert, FL.: "Tympanic plexus neuralgia. True tic douloureux of the ear or so-called geniculate ganglion neuralgia cure effected by intracranial section of the glossopharyngeal nerve." J.A.M.A., 100:1744-1746, 1933.

Reichert, F L.: "Neuralgias of the glossopharyngeal nerve with particular reference to the sensory, gustatory and secretory functions of the nerve." Arch. Neurol. Psychiat., 32:1030- -1037.1934.

RETINOHIPOPHYSARY SYNDROME

Syndromography

Clínic

It occurs at any age, more frequently in females. It is characterized by headache, vertigo and vision disorders accompanied by typical visual field alterations. In the fundus, narrowing of the retinal vessels, neuritis and atrophy of the optic nerve are observed.

Paraclinical examinations

Skull radiology. Alterations of the bone structures of the sella turcica with decalcification and osteolysis of the posterior clinoid processes.

Syndromogenesis and etiology

The syndromogenesis and etiology are not specified.

Literature

Lijo, Pavia J.: "retinopituitary syndrome treated by serum gonadotropin. Four new observations." Rev. Oto. Neuro. Ophthal.. 22:5-9, 1947.

Lijo, Pavia J. and M. Lis: “Benign retinopituitary syndrome. About 30 observations." Rev. Oto. Neuro. Ophthal., 24:41-45; 73-76, 1949.

SPASMODIC LAUGHTER SYNDROME

Synonymy

S. of forced laughter. S. of Homeric laughter. S. of attacks of laughter

Syndromography

Clínic

It should be considered a symptom rather than a syndrome.

It is characterized by uncontrollable laughter, disproportionate to the provoking cause, without emotion or pleasure.

Syndromogenesis and etiology

It is observed in multiple sclerosis, pseudobulbar palsy, intracranial hemorrhage and after lobotomy of the premotor cortex.

Literature

Grinker, R. R. and A. L. Sahs: Neurology. ed. 6, Springfield, Thomas, 1966.

Homer, in both Iliad and Odyssey: "And unextinguished laugher shakes the skies."

Martin, J. P.: Fits of laughter (sharm mirth) in organic cerebral disease." Brain, 73:453-64. 1950.

ROMBERG SYNDROME

Synonymy

S. de Parry-Romberg. S. of progressive trophoneurosis facialis. S. of progressive lamellar aplasia. S. of progressive facial hemiatrophy.

Syndromography

Clinic

Both sexes are affected and it begins in childhood. Sometimes it is preceded by trauma or trigeminal neuralgia. The onset is on the left side (62%) and is characterized by progressive atrophy of skin, subcutaneous cellular tissue, muscles, bones and cartilage; The ear becomes smaller and separated from the head, then it affects the eyebrow, corner of the mouth and neck, exceptionally the hemibody. In the injured area, the skin becomes pigmented, sometimes associated with vitiligo; There is circumscribed alopecia that also affects the eyelashes and inner half of the eyebrow. Nervous system: trigeminal neuralgia, paresthesias and Jacksonian-type seizures with ipsilateral or contralateral cerebral hemiatrophy. Eyes: enophthalmose antimongolic inclination of the eye, with extrinsic paralysis, lagophthalmos and ptosis; sometimes Horner syndrome. Inflammatory lesions (keratitis, iritis, iridocyclitis,) and cataract. Mouth: hemiatrophy of the upper lip (exposing the teeth) and tongue. In half of the cases the ascending branch of the maxilla participates.

The process may be bilateral in 5-10% of patients.

Paraclinical examinations

Biopsy and electroencephalogram. They are useful to establish the diagnosis.

Syndromogenesis and etiology

The presence of antinuclear antibodies in the serum of affected people suggests that this syndrome may be a form of localized scleroderma.

Most cases are sporadic, but some occur in families and are transmitted by autosomal inheritance.

dominant with variable penetrance, although this is not confirmed.

The etiology is unknown.

Literature

Parry, CH.: Collections from unpublished papers. London, 1825.
Romberg, M. H.: Trophoneurosen. Klinische Ergebnisse. A. Forstner, Berlin, 1846. p.75.
Schanall, B. S. and D. W. Smith: “Nonramdon laterality of malformations in paired structures." J. Pediatr., 85:509, 1974.

ROSS SYNDROME

Synonymy

S. of selective and progressive pseudomotor denervation. S. Holmes-Adie syndrome with segmental hypohidrosis.

Syndromnography

Clinic

The age of appearance is variable. It is characterized by tonic mydriasis of the pupil, decreased deep tendon reflexes, progressive hypohidrosis and anhidrosis that follows the pattern of the corresponding dermatome.

Paraclinical examinations

Sweat test. Localized hypohidrosis (pilocarpine test).

Special tests. To specify the syndrome of pupilotonic pseudotabes or Adie syndrome.

Syndromogenesis and etiology

The coexistence of hypohidrosis and Holmes-Adie syndrome does not appear to be coincidental (see Holmes-Adie syndrome).

The etiology is unknown.

Literature

Ross, A.T.: "Progressive selective sudomotor denervation; a case with coexisting Adie's syndrome." Neurology, 8:809-817, 1959.
Lucy, D.D.; M. W. Van Allen and H. S. Thompson: "Homes- -Adie's syndrome with segmental hypohidrosis." Neurology, 17:763-769, 1967.

ROUSSY-CORNIL SYNDROME

Synonymy

Adult sporadic hypertrophic neuropathy. Interstitial hypertrophic neuritis. Progressive hypertrophic interstitial neuritis.

Syndromography

Clinic

It occurs between the second and third decades of life. It is characterized by widely distributed lancinating pain (not constant), peripheral paresis, fasciculation, visual disorders and ataxia. The nerves are sometimes palpable and sensitive to the touch.

Physical examination. There is peripheral atrophy, decreased or abolition of tendon reflexes, miosis, inactive pupils, and scoliosis. Roussy-Cornil syndrome is progressive with periods of remission and exacerbation.

Paraclinical examinations

Laboratory tests. Negatives.
Cerebrospinal fluid. Occasionally increased protein.

Sensory nerve biopsy. Uniform thickening of the nerve trunks, hypertrophy of the Schwann sheath that presents a laminated, onion leaf appearance.

Syndromogenesis and etiology

This syndrome is considered a form of Von Recklinghausen neurofibromatosis. It must be differentiated from Charchot-Marie-Tooth syndrome, leprosy, polyneuritis, amyloidosis and collagen diseases. It can also be considered adult-type Déjerine-Sottas syndrome or progressive hypertrophic Déjerine-Sottas neuritis.

The etiology is unknown.

Literature

Dyck, P. J. and E. H. Lambert: "Lower motor and primary sensory neuron diseases with peroneal muscular atrophy." Arch. Neuro., 18:603-618, 1968.

Roussy, G. et L. Cornil: "Névrite hypertrophic progressive non-familial de ládulte." Ann. Med., 6:296-305, 1919.

ROUSSY-LEVY SYNDROME

Synonymy

Hereditary ataxia with muscle atrophy. S. claw foot with absence of tendon reflexes.

Syndromnography

Clinic

Appears during childhood. The child has difficulty walking and instability when standing, fainting and trembling of the hands, mental deficiency. The limbs, in addition to being ataxic, show marked weakness.

Physical examination. Shortening of the feet (pes cavus) with shrinkage of the toes, tendon areflexia, kyphoscoliosis. The prognosis is reserved. Spontaneous remissions have been observed.
Death is the result of intercurrent illnesses.

Paraclinical examinations

Electromyogram. Low driving speed.
Lumbar puncture.

Syndromogenesis and etiology

It is a hereditary degenerative syndrome. It shows similarity and is related to Friedreich's ataxia. The disorders are related to cell loss in the dorsal root ganglia and secondary degeneration of the posterior columns and myelocerebellar tracts of the spinal cord.

The etiology is genetic and is transmitted by autosomal dominant inheritance.

Literature

Roussy, G. et G. Lévy: "Sept cas dúne maladie familiale particular: "Troubles de la marche, pieds bots et arefléxia tendineuse généralisée, avec accesoirement, légere maladresse des mains." Rev. Neurol.. 33: 427-450, 1926.

SCHILDER SYNDROME

Synonymy

Diffuse periaxial encephalitis. S. de Schilder-Heubner. Diffuse inflammatory cerebral sclerosis.

Syndromography

Clínic

It is a demyelinating syndrome that mainly affects children and young adults, and is more common in males than in females. The first symptoms are usually psychological and visual, attention deficit, apathy that progresses to stupor, irritability, multiple neurological symptoms (paraparesis and quadriparesis) and sensory disorders according to the affected area.

Rapid progression with slight remissions. Fatally, it is progressive after an average of three years from the onset of symptoms.

Paraclinical examinations

Lumbar puncture. Increased cerebrospinal fluid pressure, slight pleocytosis and moderate increase in proteins.

Syndromogenesis and etiology

There is a softening of the gray and white matter of the brain. The temporal and occipital lobes are the most affected. There is swelling, fragmentation and degeneration of the axon, proliferation of microglia, fatty vacuolization and perivascular lymphocytic infiltration.

This syndrome is considered a genetic disorder with possible autosomal recessive inheritance.

Literature

Schilder, P: "Zue Kenntnis der sogenanten diffusen Sklerose (Ueber Encephalitis periaxialis diffusa)". Ztschr. F. d. ges. Neurol. or. Psychiat. Berl. or. Leipz., 10:1-60, 1912.

SCHMIDT SYNDROME (A.)

Synonymy

Vagus-accessory syndrome.

Syndromography

Clínic

It is characterized by torticollis, inability to rotate the head, language disorders and difficulty swallowing:. Reserved prognosis and in relation to the etiology.

Physical examination. Ipsilateral and unilateral paralysis of the soft palate, tongue and pharynx. Partial or total paralysis of the vocal cords, sternocleidomastoid and trapezius on the same side.

Paraclinical examinations

Radiology.

Angiography.

Lumbar puncture.

Serology. To rule out lues.

Syndromogenesis and etiology

This syndrome is frequently caused by vascular lesions that affect cranial nerves X and XI at the level of the bulb or in the jugular foramen, the point of emergence of these nerves from the cranial cavity. Less frequently it is due to infectious or neoplastic processes.

Bibliography

Schmidt, A.: "Doppelseitige Acces-sormslahmung bei Syringomyelie." Deutsch. Med. Wschr., 18:606-608, 1892.

SEITELBERGER SYNDROME

Synonymy

S. of infantile neuroaxonal dystrophy.

Syndromography

Clinic

It occurs in both sexes. After normal birth and development, difficulties in walking and maintaining posture, progressive deterioration of neurological function, and incontinence during sleep appear before the second year of life.

Physical examination. Nystagmus, strabismus, blindness. Tendon hyporeflexia or areflexia, flexion and deformities of the limbs.

Paraclinical examinations

Electroencephalogram.

Electromyogram.

Lumbar puncture.

Muscle biopsy.

Syndromogenesis and etiology

Widely disseminated lesions occur in the central nervous system with the appearance of round or oval eosinophilic structures mostly in the gray matter with swelling of axons and dendrites, degeneration of nerve cells, accumulation of fat granules in the basal nuclei of the telencephalon, cerebellar atrophy with sclerosis, degeneration of the optic pathway and spinal cord tracts.

The etiology is unknown. It is believed to be a metabolic disorder. The affected families reported in the literature are consistent with a genetic etiology that is transmitted by autosomal recessive inheritance.

Literature

Steilberger, F.: "Eine unbekannte Form von infantiler lipodspeicher kranfheit des Geshirns." Proc. Ist. Inter. Cong. Neuropath. (Rome Sept. 8-13, 1952), vol. 3, Turin, Rosenberg-Sellier, 1952, p. 323.

SYRINGOMYELIA SYNDROME

Synonymy

S. of Morvan.

Syndromography

Clinic

It appears in adolescence or young adulthood. It is a progressive myelopathy, which is characterized by presenting

Symptoms corresponding to a central syndrome of the upper cervical cord:

1. Dissociated sensory disorders: loss of thermal and pain sensitivities and preservation of touch and vibration, on the nape of the neck and upper arm (slave distribution) and later towards the hand.

2. Muscle atrophy in the neck, shoulders, arms and hands.

3. Upper thoracic kyphoscoliosis, claw hand deformity, absence of deep reflexes. Occasionally, loss of painful sensation in the face due to seizure of the descending trigeminal bundle, spasticity and ataxia, associated Horner syndrome. In advanced cases (10%), painful arthropathies (Charcot joint) develop in the shoulders, elbows and knees.

The extension of the lesion towards the medulla oblongata and rarely to the pons causes syringobulbia. Trophic disorders of the hand reach their maximum expression in this type of syringomyelia (Morvan type): trophic ulcerations and infections in the fingers.

Paraclinical examinations

Cerebrospinal fluid. Normal, only proteins are elevated when arachnoiditis is associated with tumor.

Electromyography. It reveals active and chronic denervation in the muscles of the upper limbs, but sensory nerve conduction is normal in the analgesic hand.

Nuclear magnetic resonance. It reveals the size and extension of the cavity, as well as the presence of cerebellar ectopia, arachnoiditis or intraspinal tumor and anomalies associated with development.

Syndromogenesis and etiology

This is a central cavitation of the spinal cord, which adopts an eccentric position and frequently causes unilateral long tract signs.

The mechanism is poorly understood; The cavities probably originate before birth as a dilation of the primitive epidymal canal, which remains in communication with the fourth ventricle. Cerebrospinal fluid pressure is possibly transmitted from the ventricular system to the spinal cavity, as a result of obstruction of the outlet of the fourth ventricle.

When cerebellar ectopia occurs, it can secondary cause intermittent obstruction of the subarachnoid space at the cervicomedullary junction, which causes a pressure gradient between the cavity and the subarachnoid space, especially during exercise, coughing or sneezing.

It is frequently associated with craniovertebral abnormalities, most commonly with Arnold-Chiari malformation, myelomeningoceles, platybasia, atresia of the foramen of Magendie or Dandy-Walker cysts.

The etiology is sometimes idiopathic or development-related, but may be the result of trauma, primary intramedullary tumors, extrinsic compression with central necrosis of the cord, arachnoiditis, hematomyelia, or necrotic myelitis.

Literature

Grinker, R. R. and A. L Sahs: Neurology, ed. 6, Springfield, Thomas, 1966.

Macllroy, W. J. and J. C. Richardson: "Syringomyelia: a clinical review of 75 cases." J. Can. Med. Assoc. 93:73 1, 1965.

Rossier, A. B. et al.: "Posttraumatic cervical syringomyelia." Brain, 108: 439,1985.

Sususki, M.; C. Davis; L. Symon et al.: "syringoperitoneal shunt for treatment of cord cavitation." J. Neurol. Neurosurg. Psychiat.., 48: 620, 1985.

VERTEBRA-BASILAR SYSTEM SYNDROME

Syndromography

Clinic

Occlusion of one or both vertebral arteries in the presence of good collateral circulation can occur without causing clinical or pathological damage. The most common clinical syndrome that occurs in unilateral occlusion of the vertebral artery is the lateral medullary syndrome or Wallemberg syndrome, caused by ischemia in the territory of the posterior inferior cerebellar artery and characterized by: ataxia of the limbs (taking of the cerebellar, olivocerebellar and/or spinocerebellar fibers) with fall towards the ipsilateral side, loss of taste (n. of the solitary tract VII and IX) and stiffening of the arm on the same side (due to injury to the gracilis and cuneiform tracts), Horner's syndrome (miosis, ptosis and decreased sweating), loss of thermal and pain sensitivity of the face (trigeminal injury), laryngeal and pharyngeal paralysis and soft palate and decreased pharyngeal reflex (glossopharyngeal and vagus injury) .

When the lesion occurs at the level of the basilar trunk, it causes variable degrees of infarcts of the pons, midbrain, cerebellum, occipital lobe, and medial portion of the temporal lobe.

This frequently fatal condition is characterized by the sudden establishment of a coma accompanied by bilateral paralysis of cranial nerve III, facial and lingual.

STEWART-HOLMES SYNDROME

Syndromography

Clínica

This syndrome is manifested by jerking movements of one arm (ipsilateral), irregular in their sequence and distribution, which slightly affects the contralateral limb, and is frequently associated with vertigo. It may be accompanied by other signs of cerebellar injury. The crisis lasts 2-3 min.

Paraclinical examinations

Electroencephalogram.

Angiography.

Computerized axial tomography.

Syndromogenesis and etiology

The etiology is due to a lesion in the cerebellum, of traumatic, neoplastic vascular origin.

Literature

Stewart, T.G. and G. Holmes: "Symptomatology of cerebellar tumors: a study of forty cases." Brain, 27:522-59 1, 1904.

STRACHAN-SCOTT SYNDROME

Synonymy

S. of Howes-Pallister-Landor. S. of painful neuropathy.

S. of orogenital dermatitis.

Syndromography

Clinic

This syndrome is originally recognized as Jamaican neuritis." It is characterized by disorders of the optic and peripheral nerves, which cause sensory symptoms and signs such as: painful paresthesias and hyperparesthesias of the feet, loss of superficial and deep sensitivities, and ataxia.

There is a decrease in visual acuity that can progress to total blindness and pallor of the papilla. Deafness and vertigo are rare but may be associated.

Variable degrees of stomatoglossitis, corneal degeneration and genital dermatitis occur, known as the " orogenital syndrome."

Paraclinical examinations

Blood. Hypoproteinemia, hypochromic anemia, low levels of B2 and B12 (occasionally). Decrease in transketolase activity.

Urine. Abnormal excretion of methylmalonic acid.

Syndromogenesis and etiology

There is damage to the papillo-macular bundle of the optic nerve, loss of myelinated fibers of the posterior cords of the spinal cord (gracilis or Goll bundle), and systematic degeneration of the central extensions of the sensory neurons of the lumbosacral spinal ganglia. It seems that the loss of thermal and pain sensitivity is due to axonopathy.

The etiology is of nutritional origin. It has been observed in malnourished people in Jamaica and many other tropical countries and occasionally in alcoholic patients.

The specific vitamin deficiencies that cause this syndrome have not been proven.

Literature

Adams, R. D.: "On the etiology of the alcoholic neurological diseases with special references to the role of nutrition." Am. J. Clin. Nutr., 9:379, 1961.

Strachan, H.: "On a form of multiple neuritis prevalent in the west Indies." Practitioner, London, 59:477-484, 1897.

STURGE-WEBER SYNDROME

Synonymy

Cutaneous leptomeningeal angiomatosis. Encephalotrigeminal angiomatosis. S. de Sturge-Weber-Dimitri. S. de Sturge-Kalisher- -Weber. S. de Jahnke. Kalisher S. S. de Milles-Schirmer.

Syndromography

Clinic

This syndrome is characterized by cutaneous angioma limited to one half of the face and located in the area of distribution of one or several nerves, most frequently the ophthalmic and maxillary branch of the trigeminal nerve. The angioma can extend over the mucosa of the mouth, nose and orbit, never exceeding the midline.

It is visible from birth, although at this stage it is minimal and develops later. It is winey red and hot. In addition to vascular injury, the skin may be thickened, as may the subcutaneous tissue.

The neurological manifestations may be due only to mental deficiency, but local deficit signs may also be present: hemiparesis, hemianopsia.

The first neurological symptom is usually a localized seizure. The focal neurological symptoms are crossed with respect to the side occupied by the vascular lesion.

During childhood, there is an increase in the volume of the eyeball, intraocular hypertension that produces optic atrophy and finally blindness. Exophthalmos that acquires extreme degrees. The affected hemiface may present acromegaloid alterations.

Paraclinical examinations

Simple skull radiology. Calcifications are observed in the occipitotemporal region and in the eyeball, which are arranged in sinuous, vermicular lines of unequal density. The cranial shell is thickened and at the same time dotted with less dense stippling. There is craniofacial asymmetry that is manifested radiographically by inequality in the proportions of the skeleton. The frontal and maxillary sinuses are hypertrophic and giant on the side of the angioma.

Syndromogenesis and etiology

The angioma is congenital and is due to a dysplasia of the vascular system during the first six months of fetal life that affects the vessels that surround the brain and the leptonmeninges.

There is a report of male-to-male transmission that reinforces the criterion of autosomal dominant inheritance.

Literature

Sturge, W. A.: "A case of partial epilepsy, apparently due to a lesion of one of the vaso-motor centers of the brain." Tr. Clin. Soc. London, 12:162-167, 1879; also, Br. Med. J., 1:704, 1879.

Weber, F. P.: "Right-sides, hemi-hypotrophy resulting from right-sided congenital spastic hemiplegia with a morbid condition of the left side of the brain revealed by radiogram". J. Neurol. Psychopath. London, 37:301-311, 1922.

TAPIA SYNDROME

Synonymy

S. vagus-hypoglossus. S. of the ambiguous and hypoglossal nucleus.

Syndromography

Clinic

It is characterized by dysarthria and dysphagia. There is ipsilateral paralysis of the tongue, soft palate and vocal cord with lingual hemiatrophy. Sometimes it can be accompanied by paralysis of the sternocleidomastoid and trapezius.

Paraclinical examinations

Skull radiology.

Angiography.

Computed axial tomography.

Syndromogenesis and etiology

There is paralysis of the vagus and hypoglossus (X and XII). Sometimes there may be paralysis of the external branch of the accessory nerve (XI).

This syndrome can be caused by intracranial injuries, cranial fractures, dislocation of the atlas, aneurysm or tumor lesions.

Literature

Tapia, A. G.: "Un nouveau syndrome; quelques cas d'hémiplégie du larynx et de la langue avec ou sans paralysie du aternocléido-mastoidien et du trapeze." Internat Arch. d'Laryngol., 22:780-785, 1906.

ESSENTIAL BENIGN TREMOR SYNDROME Synonymy

S. of presenile senile tremor. S. of benign tremor. S. of idiopathic, genuine, infantile-juvenile tremor.

Syndromography

Clinic

It can appear at all ages and in both sexes, although it is rare in childhood, and once it starts it lasts throughout life. In elderly patients it is called 'senile tremor'. It is characterized by the appearance of a more or less rhythmic, fine or coarse tremor that spreads progressively, which is present when muscle tone increases (static tremor) and in movement (kinetic or intentional), increased by emotions, fatigue and cold, and relieved by rest, sedation and sleep, which mainly affects the hands, arms, neck and head and sometimes the trunk, absence of parkinsonian signs (postural rigidity), good coordination, no reflexes pathological.

Syndromogenesis and etiology

Its anatomopathological basis is unknown.

Literature

Dana, C. L.: "Hereditary tremor, a hitherto undescribed form of motor neurosis." Amer. J. Med. Sci. 94:386-393, 1887.

MIDDLE BRAIN TREMOR SYNDROME

Synonymy

S. de Souquez-Bertrand. Bonhoeffer's Korean S. S. of Rubral tremor. S. of the superior cerebellar peduncles. S. of the lower red nucleus. S. of Claude.

Syndromography

Clínica

Clinical picture characterized by hemianesthesia and occasionally hemiataxia with contralateral paralysis of the eye muscles, innervated by cranial nerves III and IV.

Syndromogenesis and etiology

Occlusion (thrombosis or neoplasia) of the branches of the paramedian artery of the lower part of the red nucleus in the mesencephalic tegmentum.

Literature

Bonhoeffer, K.: Ein Beitrag zur LOkalisation der choreatischen Bewegungen." Mschr. Psychiat. Neurol., 1:6-41, 1897.

Denny-Brown, D.: The basal ganglia and their relationship to disorders of movement. London, Oxford Univ. Press, 1962.

THIEBAUT SYNDROME

Synonymy

S. of spinal cholesterinosis. (See S. de Van Bogaert-Sherer--Epstein.)

Syndromography

Clínica

It is characterized by progressive spastic paraplegia. Tendon xanthomas, hepatosplenomegaly.

Paraclinical examinations

Blood. Hyperlipemia and hypercholesterolemia.

Syndromogenesis and etiology

The etiology is unknown. It appears to be caused by a defect in lipid metabolism.

Literature

Thiebaut, F.: "Paraplégie spasmodique et xanthomes tendineux associés. Des rapports de ce syndrome avec la cholestérinose cerebrum spinale." Rev. Neurol., 74:313-315, 1942.

THOMSEN SYNDROME

Synonymy

S. of myotonia congenita. S. of dystrophic myotonia. S. of dominant congenital myotonia.

Syndromography

Clínic

It appears predominantly in males, at the time of birth or shortly after, sometimes suddenly at puberty. After an asymptomatic period, there is difficulty in muscle relaxation, frequently limited to the limbs, and in starting to walk. The chewing, laryngeal and extrinsic muscles of the eye may be affected. Emotions and cold can increase symptoms.

Sometimes cataracts develop and muscle atrophy appears in the evolution of the syndrome.

The child is unable to sit up if he is not supported and does not keep his head up. Adopt a typical posture: "frog position.

Paraclinical examinations

Muscle biopsy. It shows the typical picture of group atrophy, with hypertrophy of normal fibers.

Electromyography. Numerous fibrillation potentials, proving the denervation substrate of weakness. Decrease tion of nerve conduction velocity.

Syndromogenesis and etiology

In this syndrome there is an increase in the number of myofibrils and the amount of sarcoplasm, as well as interstitial connective tissue.

The etiology is genetic with autosomal dominant inheritance. Molecular studies excluded the possibility that

This syndrome was an allelic form of the myoclonic dystrophy gene.

At least four varieties of dominant myotonia congenita have been suggested:

Type I. Classic Thomsen syndrome.
Type II. Muscle pain and fluctuating evolution.
Type III. Cold myotonia, especially around the eyes, nose and mouth (it is a different entity from paramyotonia, since there is no cold-induced paralysis).
Type IV. It has an intermittent course without involving the facial muscles.

A possible type V is discussed due to isolated myotonia triggered by tongue percussion.

Literature

Becker, R. E.: Myotonia congenita and syndromes associated with myotonia. vol. II, Topics in Human Genetics, Stugart, Georg Thieme, 1977.

Koch, M. et al.: "Myotonia congenita (Thomsen's disease) excluded from the region of the myotonic dystrophy locus on chromosome 19." Hum. Genet., 82:163-166, 1989.

Rudel, R. and F. Lehmanm-Horn: *Menbrane changes in cells from myotonia patients." Physiol. Rev., 65:310, 1985.

Thomsen, J.: "Tonische Krampfe in wilkurlich beweglichen Muskeln in Folge vonerebter psychischerr Disposition (Ataxia muscularis ?)." Arch. Psychiat., 6:706-718, 1875-1876.

TOLOSA-HUNT SYNDROME

Synonymy

S. of the cavernous sinus.

Syndromography

Clinic

Clinical picture characterized by orbital pain, sensory loss in the distribution region of the ophthalmic branch of the trigeminal nerve and unilateral ophthalmoplegia with eyelid ptosis.

Paraclinical examinations

Orbital ultrasonography.
Computerized axial tomography.

Syndromogenesis and etiology

Aneurysm and thrombosis of the cavernous sinus, invasive tumors of the sinuses and sella turcica or at the level of the superior orbital fissure, inflammatory pseudotumor of the apex of the orbital cavity. The cranial nerves are taken: II, IV, VI and ophthalmic branch of pair V.

Literature

Kennerdell, J. S. and S. C. Dresner: "The nonspecific of orbital inflammatory syndromes." Surv. Opthalmol, 29:93, 1984.

VAGAL BODY TUMOR SYNDROME

Syndromography

Clinic

It occurs equally in both sexes. Average age of onset 37 years. Frequently asymptomatic. It is characterized due to sudden hoarseness, unilateral dull pain in the neck, syncope and dysphagia.

Physical examination. A mass is palpated in the upper lateral portion of the neck (most commonly on the right side). The peritonsillar structures are displaced medially (50% of cases), lingual hemiatrophy is rare. It may be associated with Horner syndrome.

Syndromogenesis and etiology

An encapsulated ovoid nodular mass 2-6 cm in diameter has been found below the base of the skull, near the jugular foramen. Metastases are rarely observed.

The etiology is unknown.

Literature

Oberman, H.A.; F. Holtz: L. A. Sheffer et al.: "Chemodectomas (nonchromaffin paragangliomas) of the head and neck: clinicopathologic study." Cancer, 21:838-85 1, 1968.

White, E. G.: “Die structur des glomus caroticum, seine Pathologhie und Physiologie und seine Bezihung zum Nervensystem." Beitr. Path. Anat., 96:177-227, 1935.

VAIL SYNDROME

Synonymy

S. of the vidian nerve. S. painful facial.

Syndromography

Clinic

Predominates in adult women. Generally, it is unilateral, often nocturnal. It is characterized by severe pain in the face that radiates to the nose, eye, ear, head, neck and shoulder.
It is frequently accompanied by symptoms of sinusitis. It is closely related to Sluder syndrome.

Syndromogenesis and etiology

It is due to irritation or inflammation of the vidian nerve secondary to infectious processes of the sphenoid sinus.

Literature

Vail, H. H.: "Vidian neuralgia, with special refernec to eye and orbital pain in suppuration of petrous apex." Ann. Otol. Rhine. & Laryng., 41:837-856, 1932.

VERNET SYNDROME

Synonymy

S. of the jugular foramen. S. of the posterior torn hole.

Syndromography

Clinic

It is characterized by swallowing disorders, ageusia of the posterior third of the tongue, velopalatine and laryngeal hemiplegia, hoarseness, immobility or weakness of the sternocleidomastoid and trapezius on the same side, with descent of the shoulder stump and difficulty rotating the head.

Paraclinical examinations

Skull radiology.

Computerized axial tomography.

Syndromogenesis and etiology

The symptoms are produced by paralysis of the bundle of nerves that emerge through the posterior foramen lacerum or jugular foramen (glossopharyngeal, vagus and accessory).

The most common causes are trauma, skull base fractures, and intracranial tumors of the posterior cranial fossa. It can also be caused by aneurysms.

Glomus jugulare tumor is rare.

Literature

Diamon, S. and D. J. Dalessio: The practicing physician's approach to headache (4th ed.), Butterworth, London, 1982.

Vernet, M.: Les paralysies laryngées associés. Lyon, p. 233, 1916 (Thesis).

Vernet, M.: "The classification of syndromes associated laryngeal paralysis." Med. Rev. of Rev. N.Y., pp.:449-458, 1918.

VILLARET SYNDROME

Synonymy

S. of the retroparotid region.

Syndromography

Clinic

It constitutes the exocranial variety of posterior torn condylar syndrome or Sicart and Collet syndrome.

It is characterized by ipsilateral paralysis of the soft palate, pharynx and vocal cords associated with decreased eyelid opening, enophthalmia and miosis (Bernard-Horner syndrome).

Paraclinical examinations

Eraneus radiology.

Computerized axial tomography.

Syndromogenesis and etiology

The symptoms are due to paralysis of the last four cranial nerves IX,

The causes that can cause this syndrome are:

trauma, parotid tumors and neoplasms of the retroparotid space.

Literature

Villaret, M.: “Le syndrome nerveux de léspace rétro-parotidien postérieur." Rev. Neurol., 23:188-190, 1916.

VON HIPPEL-LINDAU SYNDROME

Synonymy

Cerebellar and retinal angiomatosis. Retinoencephalic angiomatosis. S. de Lagleyse-Von Hippel-Lindau.

Syndromography

Clínic

The syndrome begins in young adults and is characterized by vision loss, progressive cerebellar ataxia, headache, and vertigo. you This diagnosis should be considered in all patients with cerebellar tumor syndrome. Angiomas and cysts occur in the brain, predominantly in the cerebellum, spinal cord, liver, pancreas and kidneys, the latter being the cause of death in some cases.

Physical examination. The fundus examination demonstrates, in addition to papilledema due to intracranial hypertension, angiomatous vascular lesions, with tortuous and thick vessels that cause retinal detachment and converge into true angiomas. This image is typical and diagnostic.

Some patients present pheochromocytomas.

Paraclinical examinations

Computed axial tomography and nuclear magnetic resonance. They show tumor lesions and/or cysts, mainly in the cerebellum.

Syndromogenesis and etiology

It is inherited in an autosomal dominant pattern with reduced penetrance of the gene. It is considered a disorder of the mesoderm during the development of the third month of intrauterine life. The responsible gene has been located at 3p26-25 and no genetic heterogeneity has been evident in families with and without pheochromocytomas.

Literature

Hardwig, P. and D. M. Robertson: "Von Hippel-Lindau disease" Ophthalmology, 91:263, 1984.

Lindau, A.: "Studien tiber Kleinhincysten" Acta Path. Microbiol. Scand., supp., 1:128, 1926.

Llanio, R.: "Retinoencephalic angiomatosis. Von Hippel-Lindau disease. Review of angiomatosis." Arch. Hosp. University. vol. I No.2 March-April, 1949.

Mather E. R. et al.: "Von Hippel-Lindau disease: a genetic study." J. Med. Genet. , 28:443-447, 1991.

Shapiro, W. R.: "Brain tumors." Semin. Oncol., 13:1, 1986.

Von Hippel, E.: Vostellung eines Paienten mit einen sehr ungewohnlichen Aderhautleiden. Bericht u.d. 24 Versammlung der Ophthl. Ges., 1895. pp. 269.

VON RECKLIGHAUSEN SYNDROME

Synonymy

S. of neurofibromatosis type I. S. of pigmentary neurocutaneous polyfibromatosis. S. of multiple

Syndromography

Clinic

It is a phacomatosis neurocutaneous disease that is characterized by café-au-lait spots and fibromatous tumors of the skin. Other clinical manifestations are: scoliosis, tibial nonunion, pheochromocytomas, meningiomas, gliomas, acoustic and optic neuromas. Hypertension, mental retardation and hypoglycemia.

This syndrome begins in childhood, and worsens during puberty, pregnancy, and menopause.

Skin spots and multiple neurofibromas are combined.

The pigmented spots are irregular in shape with relatively sharp edges, their size varies from a few millimeters to several centimeters and they are brownish brown (café au lait). They are most abundant on the trunk, axilla and in the pelvic region. The tumors are usually multiple and vary in size from tiny lesions to large tumors several centimeters in diameter. Most of them are asymptomatic;

Sometimes, if they reach a large size or according to their location, they can put pressure on adjacent structures.

Acoustic nerve tumors, generally bilateral, can be expressed clinically as a cerebellopontine angle syndrome; patients who present them always lack peripheral neurocutaneous signs of neurofibromatosis.

The most notable ocular manifestations are spots of Lish of the surface of the iris, choroidal hematomas are also seen; Optic nerve tumors can cause unilateral and rarely bilateral blindness, exophthalmos, paralysis of the extrinsic muscles of the eye and eyelid lesions.

The central nervous system is significantly affected in neurofibromatosis. There are seizures (less than 10% of patients), mental retardation, and different tumors of the central nervous system appear more frequently than usual.

Musculoskeletal involvement is common: slight shortening of height and macrocephaly, kyphoscoliosis, congenital curvature of the tibia, fibula or both, which can evolve into pathological fractures and pseudoarthrosis, asymmetric and localized gigantisms.

There is a clear increase in the incidence of certain rare malignancies, such as neurofibrosarcoma, malignant schwannoma, Wilms tumor, rhabdomyosarcoma and some forms of leukemia.

Vascular lesions such as renal artery stenosis may occur, which is usually bilateral and is due to vascular dysplasia and not compression by neurofibromas.

Other vascular lesions are coarctation of the aorta, pulmonary and cerebral stenosis.

Precocious puberty and other disorders of sexual development occur more frequently than usual.

The diagnostic criteria for neurofibromatosis type I (NF-1) in the problem patient are two or more of the following manifestations:

1. Two or more café-au-lait spots more than 5 mm in diameter in postpubertal individuals.
2. Two or more neurofibromas of any type or one plexiform neurofibroma.
3. Axillary or inguinal region freckles.
4. Optic glioma.
5. Two or more Lisch nodules (iris hamartomas).
6. Bone lesions such as sphenoid dysplasia or thinning of the cortex of long bones, with or without pseudoarthrosis.
7. A first-degree relative with a diagnosis of NF-1.

3-15% of cases develop malignancy (fibrosarcomas, squamous cell carcinomas, neurofibrosarcomas, among others).

Central neurofibromatosis is characterized by acoustic neuromas and meningiomas with few lesions or neurofibromas; it constitutes another nosological entity known as neurofibromatosis type II.

Paraclinical examinations

Slit lamp examination of the iris. To detect Lisch nodules after seven years of age.

Radiology. In search of the referred skeletal injuries.

Ultrasound.

Biopsy of the nodules.

Computerized axial tomography.

Electroencephalogram.

Nuclear magnetic resonance. It shows the presence of tumors in the spinal roots, as well as intracranial tumors and hamartomas.

Syndromogenesis and etiology

It is a syndrome of genetic etiology. The mutated gene (NF-1) is transmitted as an autosomal dominant inheritance and has been mapped to chromosome 17 in ql1.2.

The gene encodes a cytoplasmic GAP-like protein that may be involved in the control of cell growth by interaction with proteins, such as those of the RAS gene product. Abnormalities of this gene can trigger clinical manifestations.

There are cases of sectoral neurofibromatosis that could be interpreted as somaic mutations.

Literature

Binet, E. F. et al: "Orbital dysplasia in neurofibromatosis." Radiology, 93:829, 1969.

Candeville, R. et al: "Lesions yasculaires diffuses dans la neurofibromatose de Recklinghausen." Ann. Pediatr., 20:561, 1973.

Carey, J.C.; J. M. Lamb and B. D. Hall: "Penetrance and variability in neurofibromatosis: a genetic study of 60 families." Original Articles Series, XV 271, 1979.

Carlson, D. H.: Neurofibromatosis of the bladder in children." Radiology, 105:401, 1972.

Ricardi, V. M.: *Von Recklinghausen neurofibromatosis." N. Engl. J. Med., 305:1617, 1981.

Von Recklinghausen, FD.: Ueber die multiplen Fibrome der Haut und ihre Beziehung zu den multiplen Neuromen. Festschr. Feier. Funfundzwanaigjahrigen Best. Path. Inst. Berlin, Berlin A. Hirschwald, 1882.

Wallace, M. et al.: “A not I linking clone detects the 17g11.2 breakpoints in two patients with von Recklighausen neurofibromatosis (NF-1) (abstract) Cytogenet." Cell Genet. 51:1101-1111, 1989.

Xu, G. et al.: "The neurofibromatosis type I gene encodes a protein related to GAP." Cell 62:599-608, 1990.

NIH Consensus Development Conference: Neurofibromatosis: conference statement. Arch. Neurol. 45:575-578, 1988.

WALLENBERG SYNDROME

Synonymy

Obstructive S. of the inferior cerebellar artery. S. of the artery of the lateral fossa of the bulb of Foixy Hillemand. Dorsolateral medullary S. S. lateral bulbar. S. of the inferior cerebellar peduncle.

Syndromography

Clinic

This syndrome is manifested by thermal and painful hemianesthesias of the trunk and contralateral limbs, direct facial hemianesthesia, ipsilateral palatopharyngolaryngeal paralysis (dysphagia, hoarseness, paralysis of the soft palate, paralysis of the vocal cords, decreased gag reflex and loss of the sense of taste). Ataxia and lateropulsion on the same side, lingual hemiatrophy and ipsilateral Claude Bernard-Horner syndrome (miosis, enophthalmos, decreased eyelid opening). There may be bilateral nystagmus that is more accentuated towards the side of the lesion, of a horizontal type and more rarely rotary, which takes on a clockwise type when the lesion is on the left and counterclockwise on the right.

Paraclinical examinations

Angiography. It allows assessing blood flow in the distal portion of the vertebral and posterior inferior cerebellar arteries.

Computerized axial tomography. It can reveal a large cerebellar infarction in the territory of the affected artery.

Nuclear magnetic resonance. It allows cerebellar infarctions to be discovered in earlier stages and, with refinement of the technique, a lateral bulbar infarction. It is feasible to obtain images of the atherothrombotic material in the vertebral and basilar arteries and determine whether these arteries are functioning or occluded, which avoids performing angiography.

Syndromogenesis and etiology

Wallenberg syndrome is due to atherothrombotic lesions of the last segment of the vertebral artery, proximal to the origin of the posteroinferior cerebellar artery, which causes infarction of the lateral part of the medulla (medulla oblongata) with or without the posteroinferior part of the cerebellum.

The affected territory includes the retrolivar gray reticular formation, through which the fibers of thermal and pain sensitivity run (gracillis and cuneiform bundles); the nucleus ambiguus of cranial nerves IX, X and XI, descending fibers and trigeminal nucleus; the nucleus of the hypoglossus or its fibers; the restiform body (inferior cerebellar peduncle) and the oculosympathetic fibers located in the gray reticular formation.

Literature

Ausmen, J. I. et al.: "Vertebrobasilar insufficiency. A rewiew." Arch. Neurol., 42:803, 1985.

Fisher, C.M. et al. "Lateral medullary infraction. The pattern of vascular occlusion." J. Neuropathol. Exp. Neurol., 20:232, 1961.

Wallenberg, A.: "Acute bulbaraffection (Embolie der Art. cerebellar post, inf. synstr.)." Arch. f. Psychiat., 27:504-540, 1895.

WEBER SYNDROME

Synonymy

S. anterior peduncular or foot of the peduncle. St. of Leyden. S. of alternating superior hemiplegia. S. de Weber-Gubler. S. of alternating oculomotor paralysis. S. of alternating peduncular hemiplegia.

Syndromography

Clínic

This syndrome is the most common of the alternating hemiplegias. It is characterized by complete contralateral hemiplegia with paralysis of the lower half of the contralateral hemiface, associated with ipsilateral oculomotor paralysis that is manifested by eyelid ptosis, dilated fixed pupil (iridoplegia), divergent strabismus and inability to move the eyeball up, down or inward, paralysis of accommodation (cycloplegia).

Paraclinical examinations

Arteriography.

Computerized axial tomography.

Nuclear magnetic resonance.

Syndromogenesis and etiology

Weber syndrome is caused by hemorrhages or thrombosis of the central part of the midbrain (midbrain) and by tumors.

These lesions produce direct paralysis of the oculomotor fibers and the pyramidal tract, as they travel through the foot of the cerebral peduncle.

Literature

Brodal, A.: The cranial nerves in neurological anatomy in relation to clinical medicine. ed. 3. Oxford, New York. 1980.

Weber, H.: “A contribution to the pathology of the crura cerebri." Med. Chir. Tr. London, 46:121-139, 1863.

WERDNIG-HOFFMANN SYNDROME

Synonymy

S. de Hoffmann. S. of infantile muscular atrophy. S. of familial spinal atrophy. S. hereditary progressive spinal muscular atrophy.

Syndromography

Clinic

The onset of the disease is variable; It frequently occurs in the last months of intrauterine life (congenital form). On other occasions the symptoms begin in the first year of life (infantile form).

There is progressive paralysis (within 3 months), in the congenital form, the child is born already paralyzed and it is not uncommon for the mother during pregnancy to notice that fetal movements decrease to disappear at the end of the pregnancy.

There is symmetrical flaccid paralysis that begins with the muscles of the pelvic girdle and the dorsal region, followed by the scapulohumeral, arms, forearms and legs. Generalized hypotonia is observed with special softness of the muscles and great flexibility that allows the patient to be placed in the most unlikely positions. The child adopts a characteristic posture:
Upper limbs in extension, arms in abduction, internal rotation and semiflexed elbows, lower limbs in external rotation with feet in varoequinus.

When there is seizure of the cervical muscles, the child cannot raise his head. Paralytic kyphoscoliosis is often present.

The respiratory muscles are frequently interested, especially the intercostal muscles with preservation (70% of cases) of the diaphragm, which facilitates pulmonary complications that can cause death.

Paralysis of the tongue is less common and sometimes there is paralysis of the mimic muscles.

The patient's intellectual development is normal. There is abolition of tendon reflexes, but there are no pyramidal signs or sensitivity disorders.

Paraclinical examinations

Electromyography. It reveals the existence, with the muscle at rest, of fibrillation and fasciculation potentials, and upon exertion, the decrease in the number of motor units and characteristic signs of involvement of the anterior horn.

Muscle biopsy. Shows denervation patterns.

Syndromogenesis and etiology

This syndrome has a genetic etiology and shows autosomal recessive inheritance. Three types are described based on the age of onset, muscular activity and age of death.

Type I. Werdnig-Hoffmann.

Type II. Intermediate form.

Type III. Wohlfart-Kugelberg-Welander disease.

All three types have their genes on 5q. Type I IE has been localized by molecular studies, including molecular cytogenetics, at 5q12-13.3.

The basic defect is not known.

Literature

Bouwesma, G. and N. J. Lerchot: "Unusual pedigree patterns in semen families with spinal muscular atrophy; further evidence for the allelic model hypothesis." Clin. Genet., 30:145-149, 1989.

Mattei, M. G. et al.: "In situ hybridization of two markers closely flanking the spinal muscular atrophy gene to 5q12-q13.3." Cytogenet. Cell. Genet., 57:1 12-113, 1991.

Mitsumoto, H.; L. S. Adelmann and H. C. Liu: "A case of congenital Werdnig-Hoffmann disease with glial bundles in spinal roots." Ann. Neurol., 11:214-216, 1987.

Werdnig, G: "Zwei fruhinfantile hereditaire Falle von progressive muskelatrophie unter dem Bilde der Dystrophie aber auf neurotischer Grundlage." Arch. Psychiat. Nervenkr., 22:437-480, 1891.

WERNICKE SYNDROME (K)

Synonymy

S. de Wernicke-Korsakoff. Wernicke hemorrhagic polyencephalopathy. S. de Gayet-Wernicke. S. of cerebral beriberi. Acute superior hemorrhagic polyencephalitis or Wernicke pseudoencephalitis.

Syndromography

Clínic

This syndrome occurs especially in liquor drinkers, heavy alcohol users, and less often in patients with chronic gastropathies or cancer cachexias.

It usually begins with drowsiness or a tendency to clouding of the sensorium, headache, and some patients sleep for a long time until they reach coma.

One of the most striking abnormalities are ocular motor signs: bilateral cranial nerve VI palsy (external ocular motor or abducens nerve), which is accompanied by horizontal diplopia; convergent strabismus and nystagmus, which can be of three types: conjugate, vertical or horizontal (the most common);
on rare occasions primary position up or down with ascillopsia, asymmetrical horizontal caused by fixed gaze.

Ataxia affects posture and gait and can lead to prostration. The scanned word occurs in isolated cases.
There are various forms of mental deterioration: a global state of confusional apathy and disproportionate memory disorders (Korsakoff' s amnesic state) and symptoms of alcohol withdrawal, whether delirium tremens or some of its variants.

Symptoms can appear simultaneously and acutely, but it is more common for ophthalmoplegia, ataxia, or both, to precede mental symptoms by a few days and sometimes by a week or more. It may be accompanied by other disorders: nutritional, polyneuropathy, heart disease.

Korsakoff psychosis is a psychological component of Wernicke-Hoffmann syndrome. Therefore, the symptom complex should be called Werdnig Syndrome when the amnesic state is not evident and Werdnig-Korsakoff Syndrome when it is possible to recognize the ocular, ataxic and amnesic symptoms.

Paraclinical examinations

Vestibular tests. Vestibular function, measured by the response to standard caloric tests, is always altered bilaterally (vestibular paresis).

Cerebrospinal fluid. Normal or slight elevation of proteins.

Blood. Increase in blood pyruvate and marked reduction in blood transketolase (thiamine-dependent enzyme, which is involved in the diversion of exose monophosphate).

Electroencephalogram. 50% of patients show wave delay in several areas.

Syndromogenesis and etiology

Nutritional deficiency is the causal factor, specifically thiamine deficiency is responsible for most of the symptoms. Acular disorders are due to a chemical abnormality and not to irreversible structural changes. Psychological symptoms seem to depend on irreversible structural changes, probably in the medial dorsal nuclei.

It has been pointed out that the lesion lies in the pathways that contain monoamines and evidence has been presented that 3-methoxy-4-hydroxyphenylglycol (MHFG), the primary brain metabolite of adrenaline, is decreased in the central nervous system of patients with Korsakoff psychosis. This has led to the assumption that damage to ascending neurons containing norepinephrine in the brainstem and diencephalon may underlie amnesia. It has also been suggested that a genetic defect in transketolase may interfere with the pathogenesis of this syndrome and it has been found that transketolase from fibroblast cultures from these patients binds thiamine pyrophosphate less avidly than transketolase from control fibroblast cultures, a defect that would be insignificant with an adequate diet, but dangerous if dietary thiamine were scarce. This would explain why only a small proportion of alcoholics develop this syndrome.

Literature

Blass, J. P. and G. E. Gibson: "Abnormality of a thiamine requiring enzyme in patients with W.H.Synd." N. Engl. J. Med., 297:1367,1977.

McEnter, W. J. and R. G. Mair: “Memory enhancement in Korsakoff's phychosis by clonidine: further evidence for a noradrenergic deficit." Ann. Neurol., 7:466,1980.

Wernicke, K.: "Der aphasische symptomekomplex." Breslau, M. Cohn und Weigert, 1874.

WESTPHAL-PILTZ SYNDROME

Synonymy

Neurotonic pupillary reaction.

Syndromography

Clínica

Pupillary contraction that appears after vigorous closing of the eyes. It delays the light reaction followed by slow pupillary dilation.

Syndromogenesis and etiology

Unknown etiology.

Literature

Piltz, J.: "Das vagotonische Pupillenphanone von Somogyl." (Wiener Klin. Wschr., 19 N. R. 33), Neurol. Centralbl., Leipz., 33:112, 1914.

Westphal, A.: "Ueber ein bisclnicht beschriebene Pupillenphanomen." Nevr. Centralbl., Leipz, 18:161-164, 1899.

WILFRED-HARRIS SYNDROME

Synonymy

S. of neuralgia of the glossopharyngeal nerve.

Syndromography

Clinic

This syndrome affects both sexes, usually between 25-70 years of age. It occurs less frequently than trigeminal neuralgia, although they are sometimes associated.

It is characterized by paroxysmal appearance of pain in the throat approximately in the tonsillar fossa extended by the base of the tongue and faucial crossroads with radiation to the ear, periauricular and lateral region of the neck, exceptionally it can be limited to the ear (neuralgia of the tympanic plexus). Stimulation of the irradiation zone of the IX cranial nerve, during swallowing and tongue movements (swallowing, speaking, chewing, yawning, laughing, etc.) can trigger the triggering of pain (trigger zone).

The patient complains of a burning or burning sensation in that area with a constrictive sensation in the throat. It is usually accompanied by a slight tic or contracture of the mimicry muscles. There is no motor or sensory deficit.

Syndromogenesis and etiology

Compression of the glossopharyngeal nerve (cranial nerve XI) by intracranial or extracranial tumors of the oropharyngeal region or peritonsillar abscesses or the base of the tongue. Rarely, shingles can affect this nerve.

Literature

Brodal, A.: The cranial nerves in neurological anatomy in relation to clinical medicine. 3rd ed. Oxford, New York, 1980. pp. 440- -577.

Loeser, J.: Trigeminal and glossopharingeal neuralgia in current therapy in neurology diseases. R.T. Johnsoned. Philadelphia, B.C. Decker, 1985-1986. pp:86-89.

WYBURN-MASON SYNDROME

Synonymy

Cerebroretinal arteriovenous aneurysm.

Syndromography

Clínic

It is a rare syndrome. It occurs more frequently in males, from birth. The appearance of the

Symptoms occur gradually or suddenly in the third decade of life and are characterized by unilateral vision loss, severe headache, vomiting, exophthalmia, hemianopia and sometimes hemi-paresis.

Physical examination. Multiple, vascular, occasionally pigmented cutaneous facial nevi, usually ipsilateral in the affected eye, in the area of distribution of the trigeminal nerve. Fundus examination shows arteriovenous aneurysm and papilledema; nystagmus. Signs of intracranial hypertension. It may be associated with other congenital anomalies.

Paraclinical examinations

Computerized axial tomography.

Angiography.

Syndromogenesis and etiology

Of possible genetic etiology, family reports suggest autosomal dominant inheritance.

Literature

Wyburn-Mason, R.: “Arteriovenous aneurysm of midbrain and retina, facial naevi and mental changes." Brain, 66:163-203, 1943.

Yokohama et al.: *Familial occurrence arteriovenous malformation of the brain". J. Neuorosurg., 74:585-589, 1991.

RELATED NEUROLOGICAL SYNDROMES

Syndromes with spinal muscular atrophy

cerebrospinal muscular atrophy

Duchenne II (progressive bulbar palsy: PBP):

Childhood PBP. Fazio-Londe type.

Adult PBP.

PBP with ophthalmoplegia.

Erb-Charcot (spastic spinal paralysis).

Charcot II (amyotrophic lateral sclerosis).

Cerebellar-spinal muscular atrophy (spin-cerebellar heredoataxia)

Friedreich I (spinocerebellar ataxia).

Roussy-Lévy.

Gertsmann II.

Richards-Rundle.

Spinocerebellar ataxia with spinal muscular atrophy (Hoff).

Spinal muscular atrophy (SMA).

Werdning-Hoffmann (progressive infantile SMA):

Neonatal form or malignant subacute early childhood.

Chronic malignant infantile form.

Benign form of second childhood.

Kugerberg-Welander (pseudohypertrophic SMA of the pelvic girdle in young people and adults).

Aran-Duchenne (distal spinal muscular atrophy) aggressive).

Brossard-Kaeser (progressive scapuloperoneal SMA).

Vulpian-Bernhardt (progressive cervico-scapulohumeral SMA) siva).

Spinal muscular atrophy-progressive acrodystrophy type Dyck-Lambert.

Cerebellar syndromes

Infantile acute cerebellar ataxia.

Agenesis of the vermis.

Alper.

Alzheimer's.

Arnold-Chiari.

Bailey-Cushing.

basal ganglia.

Bourneville.

Brun.

Cerebellar aplasia.

Cerebellar catalepsy.

Cerebellarpontine angle.

Cestan-Chenais.

Hereditary congenital ataxia-cataract dwarfism-deficiency mental.

Dandy-Walker.

Familial cerebellar ataxia-hypogonadism.

Familial cerebellar cortical atrophy.

Fourth ventricle.

Friedrich's ataxia.

Guillain-Alojouanine-Bertrand-Garcin.

Hartnup.

Hertwig-Magendie.

Jackson's cerebellar crisis.

Late cerebellar cortical atrophy.

Marie's ataxia.

Marinesco-Sjögren.

May-White.

Menke II.

Multiple sclerosis.

Died.

Cerebellar-myxedematous.

Neocerebellar.

Melanosis-neurocutaneous.

Norman-Tingey.

Nothnagel.

Olivopontocerebellar atrophy.

Paine.

Myoclonus of the soft palate.

Paleocerebellar.

Pelizaeus-Merzbacher.

Pik II.

Postinfectious cerebellar ataxia.

Hunt I.

Stewart-Holmes.

Superior cerebellar peduncle.

Tay-Sachs.

Vogt-Spielmeyer.

Von Hippel-Lindau.

Wallenberg.

Wilson.

Degenerative syndromes of the nervous system

Friedreich's ataxia.

Alzheimer's.

Bassen-Könzweig.

Bourneville.

Cerebellar (parenchymal degeneration type).

Hereditary chronic progressive chorea.

Creutzfeldt-Jakob.

Arteriopathic dementia.

Senile dementia.

Demyelinating.

Devic.

Lenticular dystonia.

Myoclonic epilepsy.

Unverricht myoclonic epilepsy.

Multiple sclerosis.

Heidenhain.

Kuru.

Laurence-Moon-Biedl.

Leber.

Progressive supranuclear palsy.

Parkinson.

Pick II.

Refsum.

Syringomyelia.

Usher.

Demyelinating syndromes

Acute disseminated encephalomyelitis.

Balo.

Devic.

Familial spastic paralysis.

Leukodystrophy.

Multiple sclerosis.

Shielder.

Diencephalic syndromes

Transient Cushing.

Periodic hypothalamic discharge.

Froehlich.

Infantile brain gigantism.

Grahmann.

Hyperthermia-hyperphagia-hypothyroidism.

Ondine curse Russell.

Seip.

Epilepsy syndromes

Cursive epilepsy.

Jacksonian epilepsy.

Todd's post-epileptic paralysis

Big bad.

Petit mal.

Lenox triad.

Crisis or akinetic attacks.

Myoclonic

Little evil

Psychomotor.

Epileptic automatism.

Miscellaneous groups.

Adversive.

Sensory attacks.

Koshernikoff epilepsy.

Unverricht myoclonic epilepsy.

Reflex epilepsy.

Visceral epilepsy.

Epileptic0-endocrine.

Infantile spasm.

Lafora «body form» of myoclonus epilepsy.

Happy doll.

Morsier I.

Penfield.

Status epilepticus.

Myoclonic epileptic syndromes lafora body?"

Laurence-Moon-Biedl.

Leber.

Multiple sclerosis.

Parkinson.

Pick II.

Progressive supranuclear palsy.

Refsum.

Senile dementia.

Syringomyelia.

Unverricht myoclonic epilepsy.

Usher.

Rare hyperkinetic syndromes

Birnbaum (chronic progressive chorea with cerebellar atrophy).

Bonhoeffer.

Bremme

Dow-Von Bogaert.

Guillain-Bertrand-Lereboullet.

Read.

Muratow.

Tsiminakis.

Syndromes with leukodystrophy

Spongy leukodystrophy (Canavan).

Globoidcellular leukodystrophy (Krabbe).

Metachromatic leukodystrophy (sulfatide lipidosis):

Scholz.

Greenfield.

Austin.

Sudanophilic leukodystrophy (Pelizaeus-Merzbacher).

Fibrinoid leukodystrophy (Alexander).

Migraine syndromes

Horton (cluster headache).

Migraine due to alteration of the basilar artery.

Classic migraine.

Common migraine.

Ophthalmoplegic migraine-hemiplegia.

Sheder.

Vail.

Myoclonic syndromes

Unverricht myoclonic epilepsy.

Epileptic.

Myoclonic epileptic (lafora body form).

Infantile spasmodic.

Hunt I.

May-White.

Myoclonic.

Palatal myoclonic.

Neuralgic syndromes

Bernhardt-Roth.

Paresthetic static brachialgia.

Cluster type headache.

Costen.

Superior orbital fissure.

Fothergill.

Hunt.

Monbrun-Benisty.

Atypical facial neuralgia.

Upper laryngeal neuralgia.

Ralder's paratrigeminal.

Sluder.

Cough due to alteration of the Arnold nerve.

Vail.

Wartenberg.

Weisenburg.

Syndromes with hypertrophic neuropathy

Amyloidosis.

Déjerine-Sottas.

Marie (similar).

Diabetic polyneuropathy (see diabetic syndromes).

Roussy-Cornil.

Von Recklinghausen.

Syndromes with periodic paralysis

Hereditary periodic adynamia.

Periodic hypokalemic paralysis.

Periodic normokalemic paralysis.

Periodic thyrotoxic paralysis.

Pseudotabetic syndromes

Alcoholic neuropathy.

Arsenical polyneuropathy.

Diabetic pseudotabes.

Syndromes with pineal tumor

Pineal gland tumors give rise to neurological syndromes associated or not with symptoms of possible endocrine etiology:

Neurological due to pineal tumor.

Pellizzi.

Pineal-gonadal.

Syndromes affecting basal ganglia

Batten.

Luis's body.

Chronic progressive hereditary chorea of the striatum.

Déjerine-Roussy.

Double atetostis.

Lenticular dystonia.

Fahr.

Foerster.

Necrosis of the globus pallidus.

Hallervorden-Spatz.

Midbrain tremor.

Myoclonus.

Parkinson.

Progressive supranuclear palsy.

Pseudobulbar palsy.

Visual-auditory-red core hallucinations.

Tay-Sach.

Vogt.

Wilson.

Syndromes that cause flaccid children

Myelopathies and neuropathies:

Werdnig-Hoffmann.

Oppenheim.

Congenital benign hypotonia.

Guillain-Barré.

Poliomyelitis.

Spinal tumors.

Transverse myelopathy, traumatic or due to other causes.

Infantile polyneuritis.

Encephalopathic causes:

Mental deficiency (especially Down syndrome).

Progressive encephalopathy (especially Tay-Sachs).

Brain tumors.

Riley-Day.

Cerebral palsy.

Neuromuscular transmission:

Erb-Goldflam.

Abnormalities in the neuromuscular junction.

Myopathic causes:

Infantile muscular dystrophy.

Polymyositis.

Glycogen storage.

Central core.

Universal muscle hypoplasia.

Ligaments and tendons:

Ehlers-Danlos.

Arachnodactyly.

Congenital elasticity of ligaments.

Arthrogryposis multiplex congenita.

Osteogenic causes:

Van der Hoeve.

Ricketts.

Scurvy.

Syphilitic pseudoparalysis.

Non-muscular causes:

Malnutrition.

Vitamin deficiencies.

Chronic diseases.

Diseases due to acute infections.

Endocrinopathies (especially hypothyroidism and hypopitweetarism).

Metabolic.

CHLOROPROMAZIN SYNDROME

Syndromography

Clínica

The clinical picture is dominated by ocular manifestations caused by deposits of fine particles in the cornea and lens that produce star-shaped opacities (cataracts) in the anterior part of the lens associated with brownish, grayish or purplish discoloration of the skin exposed to the sun.

Syndromogenesis and etiology

It is caused by ingestion of products containing chlorpromazine, some of which are known by the following trade names: Largactil, Plegicil, Siquil, etc.

ELPENOR SYNDROME

Synonymy

S. of dipsomania. S. of post-alcoholic behavior.

Syndromography

Clínica

The name of this syndrome is taken from the Odyssey, in which Homer describes a similar situation that happened to a young Greek, called Elpenor.

The syndrome occurs in some people who abuse alcohol, analeptic or hallucinogenic drugs. When these individuals wake up, in an unknown environment and in a still unconscious or semi-conscious state, they present abnormal behaviors of an antisocial or dangerous nature for themselves and others.

Syndromogenesis and etiology

It is due to alcohol poisoning or use of other drugs.

Literature

Carrot, E.; J. Velluz and Rigal: "Syndrome d'Elpenor". Presse Med., 55:573, 1947.

Rees, Ennis: The Odyssey of Homer (newly translated for the modern reader). New York, Randon House, 1960. p. 171.

OCCUPATIONAL EPISTAXIS SYNDROME

Synonymy

Occupational epidemic.

Syndromography

Clinic

There is moderate rhinorrhea and conjunctival irritation. Hemorrhages occur in patients who work in apple packing and processing where aniline dyes are used.

Syndromogenesis and etiology

Irritation of the nasal mucosa due to aniline dyes.

Literature

Quinby, G.E.: "Epidemic nosebleeds in apple packers." JAMA, 197:165-168, 1966.

Quinby, GE.: "Gentian violet as a cause of epidemic occupational nosebleeds." Arch. Environ. Health, 16:485-489, 1968.

GRAY SYNDROME

Synonymy

Neonatal toxicity due to chloramphenicol. S. gray of the newborn

born.

Syndromography

Clinic

It is characterized by the appearance of vomiting, dyspnea and insufficiency to breastfeed. The skin takes on a grayish color, and there is cyanosis. Sometimes there is jaundice, muscle flaccidity, abdominal distention, tachycardia, hypotension and cardio- collapsing.

vascular.

Paraclinical examinations

Blood. Hyperbilirubinemia.

Stool. Grayish color.

Syndromogenesis and etiology

It is considered to be caused by a failure in the hepatic metabolism and renal excretion of chloramphenicol, probably due to enzymatic immaturity in the fetus and newborn.

Literature

Burns, L.E.; J.E. Hodgman and A.B. Cass: "Fatal circulatory collapse in premature infants receiving chloramphenicol." N. Engl. J.

Med. 261:1318-1321. 1959.

Hodgman, J.E.: "Chloramphenicol." Pediat. Clin. N Amer..

8:1027-1042. 1961.

SMOKE SYNDROME
OF POLYMERS

Syndromography

Clinic

A few hours after exposure to hot polymer fumes, a severe cold, stiffness, tremors of the limbs, headache and sore throat occur.

Physical examination. No or moderate redness of the throat is observed. This condition disappears in a few hours, if the patient is removed from exposure.

Syndromogenesis and etiology

The syndrome is caused by inhalation of polytetrafluoroethylene fumes. It has been observed that particles of this polymer can contaminate the cigarettes of smokers who handle this substance, which can also cause the clinical picture.

Literature

Barnes, R.; A.T. Jones: "Polymer-fume fever. Med. J. Australia, 2:60-61, 1967.

Harris, D.K.: "Polymer-fume fever." Lancet, 2:1008-1011, 1951.

HYDANTOIN INGESTION SYNDROME

Synonymy

S. of hypersensitivity to anticonvulsants.

Syndromography

Clinic

The condition observed generally consists of a skin eruption of different types: morbilliform, scarlatiniform, urticarial or exfoliative, associated with fever and lymphadenopathy, especially in the cervical region and hepatosplenomegaly. This clinical picture begins between 4-6 weeks after continued ingestion of anticonvulsants.

sivants.

It is relatively common to see side effects in patients who take this drug permanently: gingival hyperplasia, lymphadenopathy, hirsutism and/ or cerebellar syndrome, especially if the doses are high.

Paraclinical examinations

Blood. Eosinophilia.

Liver tests. Aspartate aminotransferase and alanine aminotransferase, elevated.

Syndromogenesis and etiology

Ingestion of hydantoin in susceptible patients. Some of the commercial preparations that contain hydantoin and its derivatives are: Antisacer, Comital, Dantoinal, Dantonil, Epamin, Mesantoin, Om-Hidantin, Composite Om-Hydantoin, Sedantoinal, Vincidol and Neovincidol.

A metabolic alteration has also been considered, since this drug is metabolized in the liver by the cytochrome P450 system. When there are congenital or acquired enzyme deficiencies that have the function of eliminating the intermediate metabolites of this substance, the disorder would become more serious.

Mortality can reach up to 40%.

HYDRALAZINE INGESTION SYNDROME

Syndromography

Clinic

Ingestion and/or poisoning by hydralazine generally produces a clinical picture similar to systemic lupus erythematosus, with a predominance of joint symptoms.

Paraclinical Examinations

Blood. LE positive cells.

Syndromogenesis and etiology

Ingestion of hydralazine in susceptible patients. Apresolina and Nepresol are examples of commercial medications that contain this chemical compound.

INTOXICATION SYNDROME
BY SALICYLATES

Syndromography

Clínic

The condition is predominantly seen in children, rarely in adults. In children, it is clinically characterized by vomiting, fever, hyperventilation, CNS excitement, progressive lethargy, and coma. In adults, tinnitus, dizziness, hearing loss, blurred vision, sweating, facial flushing and hemorrhages are observed, and occasionally neurological symptoms such as hyperventilation, excitement, confusion, delirium, muscle twitches and progression to lethargy, stupor and coma.

Paraclinical examinations

Blood. Initially respiratory type alkalosis, hypokalemia and hypernatremia with a compensatory increase in urinary base excretion that occurs between 1 and 8 h after poisoning. An increase in lactic and pyruvic acids is also observed. Hyperglycemia, prolonged prothrombin time and frequent leukocytosis.

Urine. Oliguria, glucosuria, albuminuria and ketonuria.

Syndromogenesis and etiology

It is due to excessive ingestion of aspirin or salicylates; it can also be caused by abuse of ointments containing salicylates.

Literature

Goldberger, E.: A Primer of water, electrolyte and acid-base syndromes. ed. 3, Philadelphia, Lea & Febiger, 1963.

Singer, R.B.: "Acid-base disturbance in salicylate intoxication". Medicine, 33:1-13, 1954.

TRIPARANOL INTOXICATION SYNDROME

Syndromography

Clinic

The clinical manifestations produced by intoxication with this drug consist of alopecia and hair depigmentation, ichthyosis and cataracts.

Syndromogenesis and etiology

Triparanol poisoning.

Malignant syndrome due to neuroleptics

Synonymy

S. of malignant hyperthermia. King's S. S. of hyperthermia of anesthesia. S. of Evans myopathy (affected family). S. of malignant myopathy-hyperpyrexia.

Syndromography

Clínica

It consists of the appearance of hyperthermia, extrapyramidal rigidity and disorders of the autonomic nervous system in patients subjected to neuroleptic agents, regardless of the dose and route of administration of these substances. In patients in whom the complete clinical picture occurs, it is characterized by a rapid rise in temperature to 39-42°C, associated with rigidity and akinesia, which occurs after inhalation of anesthetics such as halothane, methoxyflurane, cyclopropane and ethyl ether, or the administration of muscle relaxants such as succinylcholine.

Dysfunction of the autonomic nervous system can be manifested by tachycardia, unstable blood pressure, profuse sweating, dyspnea and sphincter incontinence, and can persist after 5-10 days of administration of neuroleptics.

The overall mortality is 20% and can occur up to 30 days after discontinuation of neuroleptics when they are administered orally.

In 90% of individuals susceptible to this syndrome, muscles contract when exposed to concentrations of caffeine, halothane, or hexamethonium. There is a second form of malignant hyperthermia (recessive type), known as King's syndrome, which occurs in young people, less frequently in women, which is accompanied by some congenital malformations such as short stature, undescended testicles, lumbar lordosis, thoracic kyphosis, pectus carinatum, winged neck and scapulae, small chin, low set of ears and antimongoloid obliquity of the palpebral folds.

The Noonam phenotype (25%) and always familiarity (in a family 11 affected with 30 members) allow us to suspect the diagnosis before subjecting them to anesthetics or neuroleptics.

Mortality is high.

This syndrome has also been described in other severe myopathies, including congenital myopathy, central core disease, and Duchenne muscular dystrophy.

Paraclinical examinations

Blood. Leukocytosis (15,000-30,000). Metabolic acidosis, hyperkalemia and hypermagnesemia. Elevation of blood lactate and pyruvate. Elevated creatine phosphokinase, 2-10 times its normal value.

Electromyogram. Continuous polyphasic muscle activity at rest, with low amplitude and normal nerve conduction velocity.

Syndromogenesis and etiology

There is a marked similarity of this syndrome with malignant hyperthermia that makes them practically indistinguishable, which suggests that it is a hereditary problem in which there is a disorder of calcium metabolism at the level of the muscle fibers.

There would also be a lack of coupling. The theory of the existence of a blockade at the level of dopaminergic receptors in the hypothalamus and basal ganglia has also been proposed.

The etiology is genetic and is transmitted in an autosomal dominant manner. There are families in which the father and the son, or the mother and the daughter, are affected; there is a group (25%), all males, with Noonam phenotype (Turner XX-XY); According to King, there is also a dominant type with high blood enzymes and another with myotonia congenita.

Literature

Downey, GP. et al.: Neuroleptic malignant syndrome. Patient with unique clinical and physiological features". Am. J. Med., 77:338, 1984.

MILIAN SYNDROME (ARSPHENAMINE)

Synonymy

S. of erythema of the ninth day. S. of Milian erythema.

Syndromography

Clínic

It appears after ingestion of arsphenamine or some of its derivatives in certain people. Between the fifth and ninth days, a clinical picture of abrupt onset occurs with general malaise, fever, chills, anorexia, vomiting, headache and sore throat, but whose main characteristic is the appearance (frequently on the ninth day) of a generalized rash that lasts between 1 and 12 days and is followed by peeling. This rash is described as erythematous or macular and is rarely of the urticarial type. Sometimes it is accompanied by conjunctival irritation, generalized adenopathy, jaundice and hepatosplenomegaly.

The clinical picture is self-limiting and recovery is spontaneous. It is never fatal and recurrence due to new ingestions of the drug is inconsistent.

Syndromogenesis and etiology

This syndrome was frequently observed in the past, during the treatment of syphilis in the pre-penicillin era.

Its etiology is uncertain and the reaction to arsenical compounds is not related to the presence of syphilis.

It is considered a toxic reaction with skin allergy.

Literature

Milian, G.: “Arsénobenzol, érytheme et rubeole". Paris Méd., 23:131-135, 1917.

Peters, E.E.: "The syndrome of Milian's erythema of the ninth day, report of 54 cases." Amer. J. Syph., 25:527-556, 1941.

NARCOTIC WITHDRAWAL SYNDROME

Synonymy

S. of morphine withdrawal.

Syndromography

Clínica

This is a set of symptoms that are observed among addicted people, in which the abrupt suspension of the drug occurs. The characteristics of the frame depend on the

intensity of the individual's addiction. Rhinorrhea, profuse sweating and extreme restlessness are observed in low-dose addicts. In those addicted to high doses we find nervousness, irritability, continuous sneezing, profuse tearing, yawning, leg pain, anorexia, nausea, vomiting, abdominal pain and diarrhea. In people addicted to very high doses the condition is much more intense and is characterized by prostration, weak pulse, massive sweating, manic behavior and generalized muscle tremors.

Syndromogenesis and etiology

The syndrome is caused by abrupt discontinuation of morphine.

PSEUDOLYMPHOMA SYNDROME

Synonymy

S. sensitivity to anticonvulsant drugs.

Syndromography

Clinic

From one week to 2 years after treatment with anticonvulsant medications, fever, malaise, and arthralgia appear.

The patient has a feeling of intense discomfort, the fever is irregular. There are polymorphic skin eruptions, generalized lymphadenopathy, hepatosplenomegaly, increased joint volume.

Regression in 7-14 days of all symptoms, when treatment is suspended.

Paraclinical examinations

Blood. Moderate anemia, leukopenia or leukocytosis, eosinophilia (5-40%).

Glutamic oxalacetic transaminase. Elevated

Lactic dehydrogenase and alkaline phosphatase. Elevated.

Radiology. Negative.

LE cells. Negative.

Bone marrow. Eosinophilic hyperplasia.

Node biopsy. Complete loss of architecture that is replaced in its germinal center by monstrous reticuloendothelial cells, which resemble Stenberg cells.

Syndromogenesis and etiology

It is due to a sensitivity to anticonvulsants (diphenylhydantoin). Dilantin and methylphenylethylhydantoin, mesantoin, phenobarbital among the most common.

Literature

Coope, R. and R.G.R. Burrows: "Treatment of epilepsy with sodium dyphenyl hydantoinatate." Lancet, 1:490-492, 1940.

Schreiber, M.M. and J.G. MeGregor: "Pseudolymphoma syndrome. "A sensitivity to anticonvulsant drugs". Arch. Derm., 97:297- -300, 1968.

PURPLE PEOPLE SYNDROME

Syndromography

Clinic

Although initially it was thought that it only occurred in females, it is now known that it can occur in patients

Psychiatric patients of both sexes undergoing long-term treatment with chlorpromazine. A peculiar grayish purpuric pigmentation is observed in areas of the skin exposed to the sun that evolve into a permanent bluish-black or metallic gray coloration. In some patients the conea and lens may present this coloration.

Syndromogenesis and etiology

The syndrome is caused by deposits in the skin of a photosensitive metabolite of chlorpromazine.

Literature

Greiner, A.C. and K. Berry: "Skin pigmentation and corneal and lens opacities with prolonged chlorpromazine therapy." Canada Med. Ass. J., 90:663-665, 1964.

PHENOBARBITAL SENSITIVITY SYNDROME

Syndromography

Clínica

The clinical picture is characterized by fever, cutaneous erythema and skin hemorrhages (purpura) and confusional alterations in consciousness.

Paraclinical examinations

Blood. Hypoalbuminemia, hypoproteinemia and biochemical alterations.

Syndromogenesis and etiology

Sensitivity to phenobarbital.

DRUGS SYNDROMES

alcoholic syndromes

Cardiomegaly-alcoholic emphysema.

Elpenor.

Rose gardener, alcoholic.

Korsakoff.

Marchiafava-Bignami.

Murri.

Acute reversible alcoholic muscle.

Alcoholic neuropathy.

Saturday night paralysis.

Alcohol suppression.

Wernicke.

Zieve.

Syndromes with pyridoxine dependence

Xanthuremic aciduria.

Pyridoxine-dependence anemia.

Cystathioninuria.

Pyridoxine-dependence seizures.

Homocystinuria.

Narcoleptic syndromes

Catalepsy.

Hynagogic hallucinosis.

Narcolepsy.

Sleep paralysis.

SYNDROMES A AND V

Synonymy

S. vertical. S. not concomitant. S. of horizontal strabismus. S. of esotropia A. S. of exotropia A. S. of esotropia V. S. of exotropia V.

Syndromography

Clínic

(Exophoria: heterophoria, lack of parallelism between visual axes.)

Patterns have been created in an arbitrary way to classify these alterations:

1. Esotropia A, greater upward than downward.
2. Esotropia V, greater downward than upward.
3. Exotropia A, greater downward than upward.
4. Exotropia V, greater upward than downward.

These patterns correspond to the forms of presentation indicated below:

1. Esotropia A, isodeviation greater than 10 diopters in upward gaze than in direct forward gaze.
2. Esotropia V, isodeviation greater than 15 diopters in downward gaze than in direct forward gaze.
3. Exotropia A, isodeviation greater than 10 diopters in downward gaze than in direct forward gaze.
4. Exotropia V, isodeviation greater than 15 diopters in upward gaze than in direct forward gaze.

Syndromogenesis and etiology

Three variants arise in the etiology of this syndrome: damage to the horizontal muscles, damage to the muscles involved in vertical movements and combined injuries.

The etiology is unknown.

Literature

Dunlap, E.A.: “Present status of the A and V syndromes.” Amer. J. Opthalmol., 52:396-401, 1961.

Urist, M.J.: "Horizontal squint with secondary vertical deviations." A.M.A. Arch. Opthal., 46:245-267, 1951.

SLOW ACCOMMODATION SYNDROME

Syndromography

Clínic

Blurred vision (asthenopia) that appears in near vision within a few minutes of starting to read, sew, or observe objects up close.

Physical examination. Accommodation disorders. Increase in the amplitude of adduction. Abnormal relaxation of the modation, induced by the relative divergence at close distances. Latent, insufficient convergence.

Paraclinical examinations

Keep patients under observation until blurred vision appears, at which point it is easy to detect typical findings.

Syndromogenesis and etiology

Accommodation fatigue, presbyopia, hyperopia, accommodation paralysis.

Literature

Hill, R.V.: "Accommodative-effort syndrome: pathologic physiology." Amer. J. Opthal., 34:423-43 1, 1951.

ADIE SYNDROME

Synonymy

S. of pupillary myotonia. S. of pupillotonia. S. of Argyll- -Robertson pupillary with mydriasis. of Argyll-Robertson non-luetic. If it is associated with alteration of deep tendon reflexes, it is known by the names: S. Markus, S. Weill-Reys Holmes IV, S. Holmes-Adie, S. pseudotabetic pupillotonia.

This syndrome is also known as pseudotabetic syndrome with pupillotonia.

Syndromography

Clinic

Sudden deterioration of vision with blurred vision, difficulty reading and headache. More common in women in the second and third decades of life.

Physical examination. Unilateral pupillary dilation (80%), absence of accommodation or very slight and less severe compromise with response to accommodation-convergence. The pupil, after slowly constricting, can sometimes remain smaller than normal. There may be a decrease or absence of tendon reflexes. Tonic pupils are dilated with atropine. The differential diagnosis must be made with Argyll-Robertson syndrome.

Paraclinical examinations

Methacholine test, A 2.5% solution instilled into the conjunctival sac, produces rapid constriction of the miotic pupil. The pupil reacts normally to adrenaline and COcaine.

Syndromogenesis and etiology

Occasionally this syndrome is associated with post-traumatic conditions or infectious diseases.

The etiology is unknown.

Literature

Holmes, G.: "Partial iridoplegia with symptoms of other diseases of nervous system." Trans. Opthal. U.K., 51:209-228, 1931.
Lowenfeld, I.E. and H.S. Thompson: "The tonic pupil: a re-evaluation." Amer. J. Ophthal., 63:46-87, 1967.
Lowenstein, O. and ILE. Lowenfeld: Pupillotonie pseudotabes (syndrome of Markus-Weill and Reys-Holmes-Adie): a critical review of the literature". Survey-Ophthal., 10:129-185, 1965.
Markus, C.: "Notes on a peculiar pupil phenomenon in two cases of partial iridoplegia." Lancet, 2:i257, 1905.
Piltz. J.: "Ueber neue Papillenphaenome." Neur. Zentralbl.. 18:248-254, 1899.

CONGENITAL ALACRIMA SYNDROME

Syndromography

Deficiency of tear secretion.

Syndromogenesis and etiology

Congenital presentation in children is rare, as is the unilateral form. The reported cases can be classified into five categories:

1. Lack of tears in the newborn.
2. Neurogenic hyposecretion.
3. Absence or hypoplasia of the salivary glands.
4. Associated Riley-Day syndrome.
5. Associated anhidrotic ectodermal dysplasia.

Literature

Smith, R.S.; S.F. Maddox and B.E. Collins: “Congenital alacrima.” Arch. Ophthal. (Chicago) 79:45-48, 1968.
Thurnam, J.: "Two cases in which the skin, hair and teeth were very imperfectly developed." Med. Chir. Trans., 31:71-82, 1848.

ALEZZANDRINI SYNDROME

Syndromography

Clinic

It occurs in adolescents and young adults. There is unilateral vision deterioration after months or years, facial vitiligo, premature hair discoloration (poliosis) that appears on the same side. Bilateral perceptual deafness develops.

Paraclinical examinations

Studies of visual and auditory perceptions.

Syndromogenesis and etiology

It appears to be due to degenerative retinitis.
The etiology is unknown.

Literature

Alezzandrini, A.A.: "Manifestation unilaterale de degenerescence tapeto-retinienne, de vitiligo, de poliose, de cheveux blancs et d' hypoacousie." Ophthalmologica (Basel), 147:409-419, 1964.

AMALRIC SYNDROME

Synonymy

Dystrophic macular S. with deaf-muteness.

Syndromography

Clinic

It occurs in both sexXOs early in childhood around 5 years of age. There is partial deafness (50 to 60%), minor visual defects with normal visual fields, slight disorders in dark vision and color perception.

Physical examination. Foveolar redness that causes counter-te with a grayish background, pigmentation spreads sometimes towards the periphery, usually

Paraclinical examinations

Auditory and visual perception exams.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance; It can also be caused by embryopathy or postnatal infections.

The etiology is unknown.

Literature

Amalric, P.: “A new type of tapetoretinal degeneration in the course of deafmutism." Bull. French Soc. Ophthal. 73:196-212, 1960.
Remky, H.; A. Klier and J. Kobor: "Maculadystrophie bei Taubstummheit (syndrom von Amalric)". Klin. Mbl. Augenh. 114:180-187, 1964.

AMAUROSIS FUEGA SYNDROME

Syndromography

Clinic

Fleeting loss of vision. The recurrence of these crises can lead to its total loss.

Physical examination. Depending on the etiology, there may be arterial hypertension and polycythemia.

Paraclinical examinations

Fundus examination.

Angiography of the vertebrobasilar system.

Syndromogenesis and etiology

Vascular deficiency can cause retinal ischemia and cause necrosis. The causes of this ischemia can be:
smoking, high blood pressure, polycythemia, cerebral stasis, carotid insufficiency (which affects the sufficiency of the ophthalmic artery), and retinal artery obstruction due to microembolism due to atherosclerotic lesions.

Literature

Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The metabolic basis of inherited disease. ed. 2, McGraw-Hill, New York, 1966.

NUTRITIONAL AMBLYOPIA SİNDROME

Synonymy

S. of retrobulbar neuropathy. S. of Obal. Multicarriage dystrophy and optic neuritis. S. of amblyopathy from the concentration camps. S. of amblyopia.

Syndromography

Clinic

It occurs in malnourished people. The onset is slow and insidious, there is progressive loss of vision, obscuration of near or distance vision, difficulty reading, photophobia, retrobulbar discomfort.

Physical examination. Initially there is a discrete loss of color of the temporal margin of the optic disc, later pallor, central or paracentral scotoma, symmetrical and bilateral, peripheral fields intact.

Paraclinical examinations

Blood. Hypoproteinemia, hypochromic, macrocytic or microcytic anemia. Low levels of vitamin B12 in the blood. Low level of transketolase.

Urine. Abnormal excretion of methylmalonic acid.

Syndromogenesis and etiology

This syndrome is a consequence of chronic malnutrition, vitamin B12 deficiency, diabetes mellitus. It may also occur after prolonged treatment with isonicotinic acid hydrazide.

Literature

Dreyfus, PM.: "Nutritional disorders of obscure etiology." Med. Sci., 17:44-48, 1966.
Obal, A.: Nutritional amblyopia." Am. J. Ophth. 34:857-865, 1951.

ANTON SYNDROME

Synonymy

S. of visual hallucinations.

Syndromography

Clínica

Blindness, confabulation, hallucinations.

Paraclinical examinations

Exams that detect vision loss.

Angiography. Obstruction of occipital arteries.

Computerized axial tomography. Presence of tumor in the occipital region, which affects the thalamic venous connections.

Syndromogenesis and etiology

The etiology is not fully established. Sometimes there is a neurological tumor, arteriosclerosis with bilateral obstruction of the occipital arteries, lesion in the calcarine fissure or thalamic lesion.

Literature

Anton, G.: "Ueber die Selbstwahr-nehmung der Herderkrankungen des Gehirns durch den Kranken bei Rindenblindheit und Rindentaub heit". Arch. f. Psychiat., 32:86, 1899.

ARLT-DAVIDSEN SYNDROME

Syndromography

Clínica

Lamellar cataract. Dental hypoplasia. Epilepsy.

Paraclinical examinations

Eye examination.

Electroencephalogram.

Literature

Mootgat, P.: Syndromes a noms propres. J. Prélat, Paris, 1966.

ASCHER SYNDROME

Synonymy

S. of blepharoachalasia. S. double eyelid tumor. S. de Laffer-Ascher.

Syndromography

Clínica

Eyes. Blepharoachalasia is the fundamental sign, two phases are described:

1 Recurrent eyelid edema.

2. Atrophy of the skin and subcutaneous tissue of the eyelids with prolapse of the contents of the orbital cavity.

Mouth. Thickening of the lips and gums. The mucosa takes on the appearance of a double lip.

Enlargement of the thyroid gland.

Paraclinical examinations

Bloody urine. Normal.

Thyroid function. Normal, occasionally it may be altered.

Syndromogenesis and etiology

Localized skin atrophy may be present.

The etiology is unknown.

Literature

Ascher, K.W.: "Blepharochalasis mit Struma und Dopperllippe." Klin. Mbl. Augenh., 65:86, 1920.
Segal, P. and S. Joblonska: Le syndrome d'Ascher", Ann. Oculist. (Par.), 194:511-526, 1961.

AXENFELD SYNDROME

Synonymy

S. of Axenfeld anomalies. S. of Hagedom.

Syndromography

Clínic

Posterior corneal disorders. Marked increase in the Schwalbe line. When associated with other disorders of the anterior segment and juvenile glaucoma, it is called Rieger syndrome. This syndrome is frequently a component of other more complex syndromes that involve the oculodentotigital systems, or is associated with other diseases that present anomalies (Marfan syndrome, myotonic dystrophy, facial deformities).

Paraclinical examinations

Radiology. Facial and dental mass.

Eye exam.

Syndromogenesis and etiology

Compromised development with persistence and deficiency of absorption and atrophy of the uveal mesodermal tissue in the anterior chamber angle (Rieger). Deficit in the development of the anterior vitreous body.

The etiology is unknown.

Literature

Axenfeld, T.: "Embryotoxon corneae posterius." Ber. Deutsch. Ophth. Ges., 42:301, 1920.

Hagedom, A.: "Congenital anomalies of the anterior segment of the eye." Arch. Ophthal., 17:223-227, 1937.

Montes, J.G.; J.C.G. Montes: "Rieger syndrome, Axenfeld anomaly with familial juvenile glaucoma." Arch. Soc. Ophth. Hisp. Amer., 27:93-99, 1967.

AXENFELD-SCHÜRENBERG SYNDROME

Synonymy

S. of congenital cyclic oculomotor paralysis.

Syndromography

Clínic

Congenital or appearing in the first year of life. Unilateral paralysis of the third nerve, ptosis of the upper eyelid, abducted eye and mydriasis (relaxation phase), which alternates with paralysis, due to automatic contraction (after 30 s ol min) of the muscle innervated by cranial nerve III, which results in lifting of the upper eyelid, miosis and deviation of the eye (spastic phase).

Paraclinical examinations

Eye exams.

Syndromogenesis and etiology

Probable congenital etiology. It has been reported after trauma. a case

The etiology is unknown.

Literature

Latorre-Morasso, S. and J. Agular: "Axenfeld-Schürenberg disease". Arch. Soc. Oftal. Hispano Am., 1:625-632, 1942.

BITOT SYNDROME

Syndromography

Clínic

It is characterized by the presence of hemeralopia, keratomalacia, conjunctival xerosis and the presence of bright gray, triangular spots and white streaks on the conjunctivae (Bitot's spots), accompanied by general decay.

Syndromogenesis and etiology

It is caused by an alteration of the mechanism that produces retinal purpura due to vitamin A deficiency.

Bibliography

Maggiore, L.: Manual of Ophthalmology. Barcelona, - Scientist-Médica, 1953. p.59.

COATS SYNDROME

Syndromography

Clinic

It predominates in male children or adolescents (juvenile form). In the adult form (see hyperlipidemic retinitis) photophobia develops with blurred vision, usually unilateral.

Physical examination. Two groups are described:

Group I. Presence of an elevation in the lower and outer part of the retina, produced by a massive exudate, bright yellow in color and deep in location. There is no evidence of hemorrhage or vascular alterations.

Group II. Marked alterations of the small retinal vessels, recent hemorrhages, plus the retinal findings mentioned in the previous group.

Paraclinical examinations

Lipidogram. It is normal in the juvenile form. In the adult form, serum lipids are elevated, particularly cholesterol.

Sabin-Feldman coloring test. It is positive in some cases.

Syndromogenesis and etiology

In the juvenile form it is assumed that a tissue factor may be the cause of cholesterol precipitation. In the adult form, chronic inflammation and hypercholesterolemia could be the triggering factors of the syndrome.

In some cases, toxoplasmosis is considered the cause.

The etiology is unknown.

Literature

Coats, G.: "Forms of retinal disease with massive exudation." Roy. London. Ophthal. Hosp. Rep., 17:440-525, 1908.

Imre, G.: "Coats' disease and hyperlipemic retinitis." Amer. J. Ophthal., 64:726-728, 1967.

Woods, A.C. and J.R. Duke: "Coats' disease: clinical findings and plasma lipid studies." Br. J. Ophthal., 47:385-412, 1963.

COGAN I SYNDROME

Synonymy

Vestibuloauditory S. with non-syphilitic keratitis.

Syndromography

Clinic

It occurs in young adults, occasionally in older people. Sudden onset unilateral or bilateral, blurred vision, eye pain, tearing, blepharospasm, nausea, vomiting, tinnitus, vertigo and rapid development of deafness. Convulsive seizures have been reported.

Physical examination. Congestive-hemorrhagic conjunctivitis.

Paraclinical examinations

Blood. Leukocytosis and slight eosinophilia.

Syndromogenesis and etiology

Manifestations of this syndrome can occur during the course of polyarteritis nodosa.

The etiology is unknown.

Literature

Cogan, D.G.: "Syndrome of nonsyphilitic interstitial keratitis and vestibuloauditory symptoms." Arch. Ophthal., 33:144-149, 1945.

Fisher, E.R. and H.R. Hellstrom: "Cogan's syndrome and systemic vascular disease: analysis of pathologic features with reference to its relationship to thromboangitis obliterans (Buerger)." Arch.
Path. (Chicago), 72:572-592, 1961.

CRYPTOPHTHALMOS SYNDROME

Synonymy

S. of cyclopismo. S. de Ullrich-Feichtiger. S. de Meyers- -Schwickerath.

Syndromography

Clinic

Cryptophthalmos (hidden eye): marked defect in the development of the eyelids and their folds. It can be bilateral, unilateral, or cryptophthalmos on one side and microphthalmos on the other side.

Other associated alterations include: malformations in the ear (middle and external), in the palatine vault. Deformity of the larynx, hoarse voice, separation of the pubic symphysis, displacement of the navel and nipples. Malformations in the fingers, meningoencephalocele, anal stenosis, congenital heart disease, renal hypoplasia or agenesis. Masculinization of the external genitalia in females. Cryptorchidism, small penis and hypospadias in men.

Paraclinical examinations

Chromosome study. It reveals the absence of the third chromosome and an additional chromosome in group C.

These alterations sometimes occur in a state of relative health.

Endocrine studies.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance. Probable alteration in the pattern of sex chromosomes.

The etiology is unknown.

Literature

Ellis, R.: "On a rare form of twin monstrosity." Trans. Obstet.
Soc., 7:160-164, 1865.

Frazer, G.R.: *XX Chromosomes and renal agenesis". Lancet, 1:1427, 1966.

Gupta, S.P. and R.C. Saxena: "Cryptophthalmuos." Br. J. Ophthal., 46: 629-632, 1962.

Pfitzer, P. and H. Muntefering: "Cyclopism as an hereditary malformation." Nature, 217:1071-1072, 1968.

Zbhender, W.: "Eine Missgeburt mit hautuberwachsenen Augen oder Kryptophthalnus." Klin. Mb. Augenheilk, 10:225, 1872.

DEJANS SYNDROME

Synonymy

S. of the floor of the orbit.

Syndromography

Clínic

Intense pain in the region of the upper jaw, hypoesthesia and paresthesia in the area of the first and second branches of the trigeminal nerve. Exophthalmos and diplopia.

Paraclinical examinations

Skull radiology.

Bacteriological examinations.

Neural studies of electrical stimulation.

Syndromogenesis and etiology

It is due to septic, inflammatory or tumor lesions that settle on the floor of the orbit.

Literature

Dejans, M.C.: "Le syndrome du plancher de lórbite." Bul. Mem.
French Soc. Ophth., 48:473-485, 1935.

Geeraets, WJ.: Ocular Syndromes. Philadelphia, Lea & Febiger, 1965.

ACQUIRED CONE DYSFUNCTION SYNDROME

Syndromography

Clínica

It appears between the fourth and eighth decades in normal individuals. The onset is insidious and is characterized by disorders in color vision, which evolves to the complete impossibility of color perception, photophobia, reduced visual acuity.

Physical examination. Flattened cones with absence of dark adaptation, decreased critical blinking frequency.

Paraclinical examinations

Electroretinogram. Abnormal.

Reading the booklet. If the patient is subjected to this type of reading in a room with normal light and then with reduced light, the patient affected by this syndrome will increase the ability to read by one line, while other patients in similar conditions will see their ability to read by one line reduced.

Syndromogenesis and etiology

Only one case with family history has been reported.

The etiology is unknown.

Literature

Larsen, H.: "Demonstration mikroscopischer pparate von einem monochromatischen Augen." Klin. Mbl. Augenheilk, 72:1, 1924.

Zweifach, PH. and Wolf: “Acquired cone dysfunction and other photopic system diseases.” Arch. Ophthal., 79:18-21, 1968.

CONGENITAL CONE DYSFUNCTION SYNDROME

Synonymy

S. of congenital achromatopsia. S. of monochromatism of the cones.

Syndromography

Clinic

It occurs from birth. There is photophobia, decreased visual acuity from 20/50 to 20/200 or less. Color vision deficit: 1, absence of color vision; 2, reduction; 3, rod and cone dysfunction (complete color blindness).

Physical examination. Occasional nystagmus. Absence of blinking in light. Normal fundus or with some lesions not pathognomonic for this condition.

Paraclinical examinations

Electroretinogram.

Syndromogenesis and etiology

The syndrome is congenital and is linked to the male sex. It is transmitted by recessive inheritance (female carrier sex). A family with dominant transmission has been reported.

The etiology is unknown.

Literature

Larsen, H.: "Demonstration mikroscopischer Praparate von einem monochromatischen Augen." Klin. Mbl. Augenheilk, 72:1, 1924.

Zweifach, P.H. andE. Wolf: "Acquired cone dysfunction and other photopic system diseases." Arch. Ophthal., 79:18-21, 1968.

HEREDITARY NON-PROGRESSIVE DEEP CORNEAL DYSTROPHY SYNDROME

Synonymy

S. hereditary polymorphic deep corneal degeneration.

Syndromography

Clinic

It occurs in both sexes, with little or no decrease in visual acuity. It is not progressive or very slowly progressive. In rare cases it is associated with glaucoma.

Physical examination. Tenuous corneal capacity.

Paraclinical examinations

Taking eye tension.

Corneal examination.

Slit lamp examination.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. The pathological lesion is limited to Descemet's membrane and the endothelium, and presents a certain opacity between the small nodular lesions.

The etiology is unknown.

Literature

Koeppe, L.: "Klinische Beobachtungen mit der Nernstspaltlampe und dem Hornhautmikroskop." Arch. Ophthal., 99:1-15, 1919.

Rubenstein, R.A. and J.J. Silverman: "Hereditary deep dystrophy of the cornea: associated with glaucoma and ruptures in Descemet's membrane." Arch. Ophthal., 79:123-126, 1968.

DOYNE SYNDROME

Synonymy

S. of honeycomb retinal degeneration.

Syndromography

Clinic

It occurs in both sexes, usually in the third decade of life, with progressive visual deterioration in 75% of cases.

Physical examination. In the fundus of the eye, spots are observed in pericapillary areas; generally the macular area is undamaged. Between the fourth and fifth decades of life, the lesions multiply and become confluent, constituting whitish atrophic-type areas that involve the optic disc and even the macular area.

Paraclinical examinations

Eye exam.

Fundus of the eye.

Syndromogenesis and etiology

It is a hereditary syndrome of autosomal dominant transmission.

The etiology is unknown.

Literature

Doyne, R.W.: “Peculiar condition of choroiditis occurring in several members of the same family.” Trans. Ophthal. Soc. U.K. 19:71, 1899.

Pearce, W.G.: *Doyne's honey-comb retinal degeneration". Br. J. Ophthal.. 52:73-78, 1968.

DUANE SYNDROME

Synonymy

S. of retraction. S. de Stilling. S. de Stilling-Türk- -Duane. S. of Türk-Stilling.

Syndromography

Clínica

Deficiency in convergence and occasionally upward or downward deviation of adduction. Reduction or loss of abduction of the eyeball, retraction on adduction with limitation of this movement, narrowing of the palpebral fissure in adduction and widening in abduction.

Paraclinical examinations

Electromyography. Alteration of the normal pattern of ocular muscles.

Syndromogenesis and etiology

Congenital defect with probable injury to the supranuclear ocular system, which determines a paradoxical synergism of the external and internal rectus muscles and their antagonists.

The etiological hypothesis is based on the electromyographic characteristics.

Literature

Duane, A.: "Congenital deficiency of abduction, associated with impairment of adduction, ratraction movements, contraction of palpebral fissure and oblique movements of the eye." Arch. Ophthal., 34:133-159, 1905.

Türk, S.: "Ueber Retractions-bewegungen der Augen." Deutsch. Med. Wschr., 22:199, 1896.

Zauberman, H.; A. Magora and J. Chaco: “An electromyographic evaluation of the retraction syndrome". Amer. J. Ophthal, 64:1103-1108, 1967.

CONGENITAL EPIBLEPHARO SYNDROME - INFERIOR OBLIQUE DEFICIENCY

Syndromography

Clínica

It is observed in childhood and is characterized by: 1, bilateral or unilateral epiblepharos, mild corneal irritation and sometimes keratitis; 2, slight or no deviation of the eye except in the field of action of the affected inferior oblique muscle, always unilateral. Contraction deficit of antagonists.

Paraclinical examinations

Eye exam.

Syndromogenesis and etiology

Queer. Only 4 cases have been reported out of 5,000 examined at the University of Obregón.

The etiology is unknown.

Literature

Swan, K.: "The syndrome of congenital epiblepharon and inferior oblique insufficiency." Am. J. Ophthal., 39:130-136, 1955.

INVERSE EPICANTOS-BLEPHAROPHIMOSIS-PTOSIS SYNDROME

PALPEBRAL

Synonymy

S. of inverse epicanthus. S. of blepharophimosis-ptosis.

Syndromography

Clinic

Bilateral eyelid ptosis, epicanthus inverse with telecanthus, eyelid phimosis or blepharophimosis, deficiency of the tissues of the upper and lower eyelids, elevation of the eyelashes, absence of eyebrows, poorly developed nasal bridge.

Paraclinical examinations

Eye exam.

Syndromogenesis and etiology

In some cases hereditary influence has been demonstrated.

The etiology is unknown.

Literature

Blair, V.P.: JB. Brown and W.G Hamm: "Correction of ptosis and epicanthus." Arch. Ophthal., 7:831-846, 1932.

Lewis, S.R.; M.S. Arons; J.B. Lynch and TG Blocker: "The congenital eyelid syndrome." Plastic Reconst. Surg., 39:271-277, 1967.

ESPILDORA-LUQUE SYNDROME

Synonymy

S. silviano-ophthalmico. S. of amaurosis-hemiplegia.

Syndromography

Clínica

Unilateral blindness and temporal contralateral hemiplegia.

Paraclinical examinations

Fundus examination.

Syndromogenesis and etiology

It is due to embolism of the ophthalmic artery with reflex spasm of the middle cerebral artery.

Literature

Espildora-Luque, C.: "Ophthalmic-sylvian syndrome." Opth Arch. Hispano-Amer., 34:616-621, 1934.

Geeraets, WS.: Ocular Syndromes. Philadelphia, Lea & Febiger, 1965.

PHOTOPHTHALMIA SYNDROME

Synonymy

S. of snow blindness. S. of ultraviolet keratitis. Electrical ophthalmic S. S. of desert blindness.

Syndromnography

Clínic

It occurs 4-5 hours after exposure to ultraviolet rays and is manifested by severe burning eye pain, intense itching, stinging of the eyelids, photophobia, tearing, blepharospasm, and vision of luminous halos around lights.

Physical examination. Miosis, congestion and edema of the conjunctiva, corneal ulceration and mucopurulent discharge.

Paraclinical examinations Eye examination.

Syndromogenesis and etiology

The injury can be caused by ultraviolet rays, snow reflections, or the luminous arc of electric welding.

Literature

Newell, EW.: Ophthalmology. Principles and Concepts. St. Louis, Mosby, 1965.

FRANCESCHETTI-KLEIN SYNDROME

Synonymy

S. of the first arch. S. jaw-facial-dysostosis. S. de Berry-Franceschetti-Klein. S. of bilateral facial agenesis. S. de Franceschetti-Zwahlen. S. of multiple facial abnormalities.

S. de Treacher-Collins-Franceschetti.

Syndromography

Clínica

It becomes evident at birth due to difficulty sucking and swallowing. There is excessive mucous secretion in the mouth and episodes of cyanosis. Obliquity of the palpebral fissure of the antimongoloid type with shortness of the lower eyelids. Short and small lower jaw, abnormal bite due to mandibular difference. Palate very arched and high. Macrostomy. Malformation of the ears. Atypicality in hair growth and distribution. Incomplete forms have been described.

Paraclinical examinations

Chromosomal studies. Negatives.

Radiological studies of bone structures.

Audiometry.

Syndromogenesis and etiology

It is an autosomal dominant inherited condition. The basic defect is unknown.

Literature

Franceschetti, A. and D. Klein: "The mandibulofacial dysostosis, a new herediatry syndrome". Acta Ophthal., 27:143-224, 1949.

McKenzie, J.: "The first arch syndrome. Develop." Med. Child. Neurol., 8:55-66, 1966.

Thomson (1846) quoted Axelsson, A., I. Brolin; H. Engstrom, et al.: Dysostosis mandibulofacialis. J. Laryng. Otol., 77:575-921, 1963.

FRANCESCHETTI-THIER SYNDROME

Syndromography

Clínica

Mental retardation, multiple lipomas, corneal dystrophy.

Paraclinical examinations

Eye exam.

Determination of mental development.

Syndromogenesis and etiology

It is an autosomal recessive inherited syndrome.

The etiology is unknown.

Literature

Franceschetti, A. and C.J. Thier: Hornhaut-dystrophien bei Genodermatosen unter besonderen Berucksichtingung der Palmoplantarkeratosen. Graefe. Archive Ophthal., 162:610-670, 1961.

FRENKEL SYNDROME

Synonymy

S. traumatic of the anterior segment.

Syndromography

Clínica

It is characterized by periocular lesions, preferably in the superointernal or internal periorbital region, absence of corneal or scleral perforation, iridodiolysis or perforation of the iris, subluxation of the lens with subsequent papillary modifications, generally stationary traumatic cataract and decrease in visual acuity to a degree that does not correspond to objectively demonstrable lesions.

Physical examination. D-shaped mydriasis (a rope appearance that replaces the arch of a sector), observed in 85% of cases. Single or multiple iris dehiscence (50%), lens opacities or subluxation (60%), retrolenticular pigment particles (56%). There may be injuries in the posterior segment.

Paraclinical examinations

Slit lamp examination.

Transillumination examination of the iris.

Syndromogenesis and etiology

The clinical characteristics are due to the varied and different injuries produced by the trauma. Sometimes they appear months and even years later.

It is caused by trauma that affects the anterior part of the eyeball.

Literature

Casanovas, J.: Ophthalmology. Ed. Rocas, Barcelona, 1961l, p.370.

Davidson, M.: The minor sequelae of eye contusions." Amer. J. Ophthal., 19:757-769, 1936.

Frenkel, H.: "Sur la valeur medical-legal du syndrome traumatique du segment antérieur." Arch. d'Opht., 48:5-27, 1931.

FUCHS SYNDROME I

Synonymy

S. of blepharochalasia.

Syndromography

Clinic

It begins in youth with transient angioneurotic edema in both eyelids, almost always the upper ones. As the disease progresses, prolapse of the orbital fat and lacrimal glands and blepharoptosis occur.

Syndromogenesis and etiology

Atrophy of the eyelids, skin and subcutaneous tissue.

The etiology is unknown.

Literature

Fuchs, E.: "Ueber Blepharochasis (Erchlaffung der Lidhaut)". Wein. Klin. Wschr., 9:109, 1896.

Segal, P. and S. Jablosky: «Le syndrome d'Ascher. Ann. D'Ocul. 194:511-526, 1961.

FUCHS II SYNDROME

Synonymy

S. heterochromic.

Syndromography

Clinic

It appears in the third and fourth decades of life. It has an insidious onset, the patient in most cases is unaware of this pathology until the opacity of the vitreous humor reduces their vision.

Physical examination. Slight or marked difference between the color of the two irises. One or both eyes may be involved (occasionally). Light, chronic cyclitis, with whitish, non-confluent precipitates, are observed in the pupillary area and lower part of the comea.

Cataracts only develop at the end of the disease.

Paraclinical examinations

Eye exam.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Fuchs, E.: "Uber Komplikationen der Heterochromia." Ztschr. Angenh., 15:191-212, 1906.
Hart, C.T. and D.M. Ward: "Intraocular pressure in Fuchs' heterochromic uveitis. " Br. J. Ophthal., 51:739-743, 1967.

GOLDENHAR SYNDROME

Synonymy

S. of oculoauriculovertebral dysplasia. S. auriculoverte-bral.

Syndromography

Clínic

At birth there are hearing defects of varying degrees, light or severe with almost total loss, conductive type deafness, visual deficit including light or severe diplopia.
Difficulty feeding. Moderate mental retardation in 10% of cases.

Physical examination.

Ocular: Epibulbar dermoid tumor. Coloboma over the eyelids or eyebrows, eyelid ptosis, antimongoloid obliquity of the auricular palpebral opening.

Handset. Small ears with auricular appendages, atresia or stenosis of the orifice of the external auditory canal, which sometimes ends in a blind fistulous orifice.

Perioral. Small nose. Unilateral facial hypoplasia or hypoplasia of the ramus or condyle of the maxilla, hypoplasia of the maxilla, very deep palatine arch. Large mouth, palatal malocclusion.

Musculoskeletal. Presence of vertebral malformations, hemivertebrae, spina bifida, sclerosis, vertebral fusions, supernumerary vertebrae, hypoplastic ribs, inguinal hernia, congenital heart defects, clubbed or crooked feet.

Paraclinical examinations

Chromosomal studies. Negatives.
Radiological studies of bone structures.
Audiometry.

Syndromogenesis and etiology

There is no evidence of hereditary factors. Embryogenic malformation that involves the first and second branchial, vertebral and ocular arches.

It must be differentiated from Treache-Collins syndrome in which the defects are bilateral and there are no vertebral malformations.

The etiology is unknown.

Literature

"Von Arlt 1845" quoted by Van Duyse, D.: "Bride dermoide oculo-palpebrale et coloboma partiel de la paupiere avec remarques sur la genese de cas anomalies." Ann. Ocul., 88:101- -132, 1882.

Berkman, M.D. and M. Feingold: "Oculoauriculovertebral dysplasia (Goldenhar' s syndrome)". Oral Surg., 25:408-417, 1968.

GREIG SYNDROME

Synonymy

S. of ocular hypertelorism.

Syndromography

Cunic

A mental deficit is sometimes associated. There may be atrophy of the optic nerve in relation to eye strain.
Strabismus and paralysis of the external rectus muscle.

Physical examination. Excessive bioorbital distance. Brachycephaly. Occipital flattening and separated by a depression in the frontal prominence, elevated nose.

Paraclinical examinations

Skull radiology. Deficiency of cranial sutures.
No signs of intracranial hypertension.

Syndromogenesis and etiology

Abnormal development of the sphenoid is proposed.

The etiology is unknown.

Literature

Greig, D.M.: "Hypertelorism." Edinburgh Med. J., 31:560-593, 1924.
Meisenbach, A.E., Jr.: "Bilateral paralysis of external rectus muscle in hypertelorism: report of a case with convergent strabism." Amer. J. Ophthal., 33:83-87, 1950.

HARADA SYNDROME

This syndrome is considered part of Vogt-Koyanagi-Harada syndrome. It was initially typified by retinal separation and absence or little anterior uveal participation.

Poliosis and alopecia occasionally present in Harada syndrome have been considered invariable findings of Vogt-Koyanagi.

Literature

Harada, E.: "Clinical study of nonsuppurative choroiditis. A report of acute diffuse choroiditis." Acta Soc. Ophth. Jap., 30:356, 1926.

HEERFORDT SYNDROME

Synonymy

S. of uveoparotitis. S. of febrile uveoparotitis. S. de Besnier-Boeck-Schaumann.

Syndromography

Clínica

Bilateral uveitis, parotitis, bilateral lymphadenopathy, facial paralysis and fever.

Paraclinical examinations

Eye exam.

Chest radiology.

Syndromogenesis and etiology

It occurs in most cases as a complication of sarcoidosis. In some it can be attributed to tubercle bacillus.

Literature

Dufour, R. and A. Bourquin: “Deux cas dúveoparotidite histologiquement tuberculeuse.” Ophthalmologica, 120:50-56, 1950.

Heerfordt, C.F.: *Ueber eine Febris uveo-parotidea subchronica an der Glanula parotis und der Uvea des Auges lokalisiert und haufig mit Paresen cerebrospinaler Nerven kompliziert". Arch. f. Ophth., 70:254-273, 1909.

Theobald, G.D. and H.L. Wilder: “Heerfordt's syndrome." Trans. Amer. Academic Ophthal. Otol., 57:332-333, 1953.

HELMHOLTZ-HARRINGTON SYNDROME

Synonymy

S. of comeal opacity and cranioskeletal dysostosis.

Syndromography

Clínica

It occurs in both sexes, from birth. There is a zonal cataract and other disorders derived from oxycephaly; apical malformations: polydactyly, syndactyly, claw hand; mental retardation: hepatosplenomegaly.

Paraclinical examinations

Eye exam.

Mental development test.

Upper abdominal ultrasound.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Francois, J.: "Syndromes with congenital cataract." Amer. J. Ophthal., 52:207-238. 1961.

Helmholtz, H. and E.F.R. Harrington: "Syndrome characterized by congenital clouding of cornea and by other anomalies." Amer. J. Dis. Child., 41:793-800, 1931.

HOMONOUS HEMIANOPSIA SYNDROOME - PARACENTRAL SCOTOMA

Syndromography

Clinic

The onset is sudden and is manifested by difficulty reading and fixating on objects located near the central field of vision of the affected eye. There is no reduction in visual acuity. Homonymous scotoma that excludes the macula, maintaining the peripheral visual field in such a way that it does not affect the field that is between the scotoma and the periphery.

Paraclinical examinations

Eye exam.

Syndromogenesis and etiology

It is considered a probable vascular accident. Embolism.

Toxic? Infections?

The etiology is unknown.

Literature

Allen, T. and H. Carman: "Homonymous hemianoptic paracentral scotoma", Arch. Ophth., 20:846-849, 1938.

Wilbrand, H.: "Ueber die makularhemianopische Lesestorung und die von Monakowsche Projektion der Makula auf die Sehsphare." Klin. Monatsbl. F. Augenh., 45:1-39, 1907.

JENSEN SYNDROME

Synonymy

S. of corneal chondrodermal dystrophy. S. of dermochondrial-corneal dystrophy.

Syndromography

Clínic

It affects both sexes. The child is normal at birth, the alterations begin in the first or second year of age. They are characterized by osteochondral deformities of the hands and feet, subluxations, and tendon contractures may develop.

Concomitantly, small, bright yellow xanthomas may occur in the form of hard nodules on the skin. Later, bilateral, peripheral or central opacities appear on the cornea.

Paraclinical examinations

Blood. Not pathological.

Radiology. A bone survey must be performed.

Syndromogenesis and etiology

It is caused by a congenital condition with no established hereditary pattern.

The etiology is unknown.

Literature

Francois, J.: Dystrophye dermochondro-cornéene familiale". Ann. doccul., 182:409-422, 1949.

Jensen, E.: “Retino-choroiditis justapa pillaris.” Graefes Arch. Ophth., 69:41-48, 1909.
Reigin, R.D. and D.B. Caplan: "Corneal opacities in infancy and childhood." J. Pediat., 69:383-392, 1966.

JOHNSON SYNDROME

Synonymy

S. adhesions. S. of pseudoparalysis of the superior rectus muscle.

Syndromography

Clínic

It is observed in children between 3-5 years old or younger than 3 years old.

Rarely later. There is strabismus that simulates a paralysis of the lateral rectus muscles. There are 4 possible variants of this syndrome, depending on the adhesive anomalies that simulate paralysis of:

1. External rectum (usually bilateral).
2. Inferior oblique (theoretical).
3. Upper rectum.
4. Superior oblique with head tilt.

Paraclinical examinations

Muscle fixation test.

Syndromogenesis and etiology

Abnormal congenital development, Etiology is

Literature

Johnson, L.V.: “Adherence syndrome: pseudoparalysis of lateral or superior rectus muscles.” Arch. Ophthal., 44:870-878, 1950.

KRAUSE SYNDROME

Synonymy

S. of congenital encephalo-ophthalmic dysplasia. S. encephalo-ophthalmic.

Syndromography

Clínic

Mnesses are usually discovered after birth in premature children. Symptoms are variable, from a minimal degree of decreased vision in only one eye to complete loss of vision. Cerebral symptoms are also of variable degree, from mild symptoms of mental deficiency to cerebral agenesis.

Physical examination. Ocular signs may be found, ranging from small areas of retinal atrophy and slight anomalies of the eyes, to ptosis, anophthalmos, strabismus, glaucoma, microphthalmia, malformations of the retina, choroid and optic nerve. Microcephaly and hydrocephalus may be seen.

Paraclinical examinations

Eye exam.

Skull radiology.

Computerized axial tomography of the skull.

Syndromogenesis and etiology

Neuroectodermal congenital defect.

The etiology is unknown.

Literature

Krause, A.C.: "Congenital encephalo-ophthalmic dysplasia." Arch. Ophthal., 36:387-444, 1946.

LEBER SYNDROME

Synonymy

S. of hereditary optic atrophy.

Syndromnography

Clínic

It usually occurs in men, it has a sudden onset, generally between 15-25 years of age. There is unilateral or bilateral vision loss, primarily affecting central vision. The association with hereditary-familial ataxia is common.

Physical examination. At first the optic disc is normal or there is edema of the optic nerve head, later atrophy.
Signs of diffuse damage to the central nervous system can be found that take on varied clinical manifestations.

Paraclinical examinations

Eye exam.

Neurological exams.

Cerebrospinal fluid.

Urine culture.

Syndromogenesis and etiology

The possibility of a toxic-metabolic disorder or an abnormality in the metabolism of cyanides or related to toxins derived from smoking is raised.

The etiology is unknown.

Literature

Adams, J.H. and Wilson W. Blackwood: "Further clinical and pathological observation on Leber's optic atrophy." Brain, 89:15-26, 1966.

Leber, T.: "Beitrage zur Kenntniss der Atrophischen Veranderungen des Sehnerven nebst Bemerkungen uber die normale Structur des Nerven." Arch. Ophthal., 14: 164-176, 1868.

BENIGN LYMPHADENOSIS OF THE ORBIT SYNDROME

Syndromography

Clinic

It generally occurs between the ages of 18 and 66. It is possible that it is more common in women.
It is characterized by exophthalmia (may be absent), edema of the orbit that resembles a slow-growing, well-circumscribed tumor with little pain. It affects one or both eyes, developing symmetrically and simultaneously. There is no adhesion to the skin; Eye movements are slightly restricted, but there is no paralysis or papilledema. In one case glaucoma developed

(lymphoadenoglaucomatous syndrome). Absence of lymphadenopathy.

Paraclinical examinations

Biopsy.

Blood and medullogram. Normal.

Syndromogenesis and etiology

Localized inflammatory process of undetermined origin (individual reaction to different stimuli: trauma, insect bites, malignant tumors of another location).

There are localized manifestations of benign lymphadenosis of the skin.

Literature

Bafverstedt, B.; C. Lundmark; H. Mossberg et al.: "Benign lymphadenosis orbitae." Act. Ophthal., 34:367-376, 1956.
Orlowski, W.J. and J. Korobowicz: "The lymphadenoglaucomatous syndrome. " Amer. J. Ophthal., 52:101-106, 1961.

RETINAL SPOT SYNDROME

Syndromography

Clinic

Normal peripheral vision. Slight deficit in dark adaptation. High incidence of macular pathology. Slight abnormality in peripheral retinal function.

Paraclinical examinations

Electrooculogram. Pathological.

Angiography test with fluoroscein staining.

Pathological.

Syndromogenesis and etiology

This syndrome includes three ophthalmological pathologies:

1. Fundic flavimaculatosis (supposed autosomal recessive inheritance).
2. Fundic colloidal bodies (possible family inheritance).
3. Albipuntatus fundica (possible family inheritance).

Literature

Klien, B.A. and A.E. Krill: “Fundus flavimaculatus". Amer. J. Ophthal., 64:3-23, 1967.
Krill, A.E. and B.A. Klien: "Flecked retina syndrome." Arch. Ophthal., 74:496-508, 1965.

NORRIE SYNDROME

Synonymy

S. of congenital ocular atrophy. S. of fetal iritis.

Syndromography

Clínic

It only affects males from birth. Blindness, in some cases mental retardation, that begins at any age. Deafness of different severity that develops between the ages of 9 and 45.

Physical examination. Presence of a mass behind the lens, which later develops cataract, corneal opacification, bulbar phthisis and occasionally atrophy of the iris with synechiae.

Paraclinical examinations

Eye and ear exams.

Syndromogenesis and etiology

A sex-linked hereditary disorder is proposed, with different expressions. Blindness is a constant finding.

The etiology is unknown.

Literature

Hansen, A.C.: Noie's disease." Amer. J. Ophthal., 66:328-332, 1968.
Norie, G: "Causes of blindness in children: Twenty-five years experience of Danish Institutes for the blind." Acta Ophthal. (Kbh.), 5:357- -386, 1927.

EYE SYNDROME OF SELAR AND PARASELAR CONDITIONS

Synonymy

S. oculohypophyseal.

Syndromography

Clínic

It is characterized by bitemporal hemianopia, although in some cases binasal hemianopia is observed.

Visual acuity is normal at first, but then becomes to be reduced, even to the point of blindness.

Initially it is just an eye. In the beginning, variability in vision is very characteristic, revealing changes in the size of the neoplasia.

Physical examination. Fundus of the eye, descending atrophy of the optic nerve. Only exceptionally does papillary stasis occur.
Ocular motility: palsy of cranial nerves VI and VIII.
Position of the eyeball: sometimes moderate exophthalmos.

Literature

Magiore, L.: Manual of Ophthalmology. Scientific Editorial--Medical, Barcelona, 1953. p. 684.

OPTOCHIASMATIC ARACHNOIDITIS EYE SYNDROME

Syndromography

Clínic

It is characterized by reduced visual acuity, field alterations such as hemianopsia, unitemporal or bitemporal, superior or inferior, concentric retraction, central scotoma or nasal retraction. A characteristic element is the variability in the behavior of vision and the visual field, due to which periods of aggravation can alternate with periods of improvement.

Extraocular symptoms include: headache, drowsiness, sometimes dizziness, vomiting, etc. Sometimes there is anosmia.

Physical examination. Depending on the course of the disease, descending optic atrophy or edematous neuritis may be discovered in the fundus; other times the fundus is normal.

Paraclinical examinations

Skull radiology. No alterations of the sella turcica.

Cerebrospinal fluid. Normal.

Syndromogenesis and etiology

Traumas and surgical interventions of the sellar region, sinusitis, syphilis, meningeal reactions dependent on endosellar or parasellar formations. All of them cause meningitis of the chiasmatic region characterized by hypertrophy and neoformation of arachnoid tissue with a cystic appearance (atrophic cystic adhesive form).

Literature

Maggiore., L.: Manual of Ophthalmology. Editorial Científico-Médica, Barcelona, 1953. pp. 682-683.

TABES DORSAL EYE SYNDROME

Syndromography

Clinic

It is characterized by the presence of atrophic lesions of the iris accompanied by discoloration, papillary alterations such as: irregularity in the shape of the pupil, miosis or mydriasis and anisocoria. There is abolition of the light reflex with persistence of the accommodation reflex and convergence (Argyll-Roberston sign), usually bilateral.

Visual acuity is more or less reduced in relation to the time elapsed.

Physical examination. The ocular fundus shows primary descending atrophy, sometimes with gray color of the papilla. A slight edematous optic neuritis is very common.

Visual field. Concentric retraction, various forms of hemianopsia. Reduced eyelid, corneal and conjunctival sensitivities. It is also accompanied by alterations in muscle balance due to paresis or paralysis.

Syndromogenesis and etiology

It is due to the lesions of the tertiary period of syphilis and is part of the clinical picture in the course of tabes dorsalis.

Literature

Maggiore, L.: Manual of Ophthalmology. Editorial, Scientific-Medical, Barcelona, 1953. p. 684.

EYE TUMORS SYNDROME

BRAIN

Syndromography

Clinic

It is characterized by the presence of isolated blepharoptosis, facial paralysis, paralysis of the ocular muscles, conjugate deviation, nystagmus. Corneal anesthesia and possible hemianopsia, depending on the topography of the tumor.

Paraclinical examinations

Fundus examination. Ophthalmoscopy reflects papillary stasis that is usually only absent in tumors of the sellar region, but not always.

Syndromogenesis and etiology

It is caused by brain tumors.

Literature

Maggiore, L.: Manual of Ophthalmology. Scientific-Medical Editorial, Barcelona, 1953. p.687.

PARINAUD OCULOGLANDULAR SYNDROME

Synonymy

S. of conjunctivitis associated with preauricular lymphadenopathy. S. conjunctivo-adenopathic.

Syndromography

Clínic

Unilateral conjunctivitis accompanied by low-grade fever.

Physical examination. Granular or ulcerative conjunctivitis, preauricular adenopathy, enlarged parotid gland.

Paraclinical examinations

Bacteriological culture. It allows the detection of viruses, mycobacteria, bacteria and fungi.

Tuberculin test. To specify cat spider disease.

Kwein and mushroom test.

Complement fixation.

Conjunctival biopsy.

Syndromogenesis and etiology

This syndrome can be seen in: cat scratch disease, tularemia, tuberculosis, lymphogranuloma venereum, coccidioidiomycosis, sporotrichosis, syphilis, sarcoidosis and listeriosis.

Literature

Muller, E: “Die differential diagnosis des konjuktivoglandularen Syndrome von Parinaud". Deutsch. Med. Wschr., 80:152-154, 1955.

Parinaud, H. and T. Galezowski: "Conjonctivitie infecteuse transmise par les animaux." Ann. Ocul., 101:252-253, 1889.

Wood, T.R.: "Ocular coccidiodomycosis. Report of a case presenting as Parinaud's oculo-glandular syndrome." Amer. J. Ophthal., 64:587-590, 1967.

OPHTHALMOPATHIC SYNDROME

Synonymy

S. thyroid-pituitary.

Syndromography

Clínic

Exophthalmia is generally bilateral, but both eyes may not be equally affected. Sometimes it is one-sided. Sometimes there is paralysis of different eye muscles.

This syndrome may be associated with a thyroid condition compatible with hyperthyroidism or Basedow syndrome, which presents with slight thyrotoxicosis, euthyroidism or myxedema.

Paraclinical examinations

Study of thyroid function.

Thyroid scan.

Skull radiology.

Eye exam.

Syndromogenesis and etiology

Pituitary hypersecretion may play a role, or it may be due to increased tissue sensitivity to the exophthalmia-producing factor, which has been isolated from the pituitary thyrotropic hormone. The role of the long-term activity of the thyroid stimulating factor, which is considered different from that which causes exophthalmia, is also discussed.

Literature

Paschkis, K.E.; A.E. Rakoff; A. Cantarow et al.: Clinical Endocrinology. ed. 3, Harper & Row, New York, 1967.

Werner, S.C. and M. Spooner: “New and simple test for hyperthyroidism employing 1-triiodothyronine and 24-hour I 131 uptake method.” Bull. N.Y. Academic Med., 31:137-145, 1955.

ORBIT PSEUDOTUMOR SYNDROME

Synonymy

S. of ophthalmoplegic exophthalmia.

Syndromography

Clinic

Pain, ophthalmoplegia, vision is generally not affected, there are no sensory disorders of the first branch of the trigeminal nerve.

Physical examination. Exophthalmos, eyelid edema, chemosis.

Paraclinical examinations

Skull radiology.

Angiography.

Syndromogenesis and etiology

Probable chronic infection.

The etiology is unknown.

Literature

Ingalis, R.G.: Tumor of the orbit and allied pseudotumors. Springfield, Thomas, 1953.

Lakke, J.P.:. "Superior orbital fissure syndrome". Arch. Neuro., 7:289-300, 1962.

CONGENITAL POSTICUS CIRCUMSCRIPTUS KERATOCONUS SYNDROME

Synonymy

S. of Haney-Falls.

Syndromography

Clínic

Increased curvature located on the posterior corneal surface, corneal cloudiness, myopic astigmatism, corneal endothelial precipitate, slight hypertelorism. Wide and flattened nose, brachydactyly, pterygium colli and barrel chest.

Paraclinical examinations

Eye examination.

Syndromogenesis and etiology

Possibly this syndrome is a type of autosomal dominant inheritance or inherited recessive defect.

The etiology is unknown.

Literature

Haney, W.P. and H.F., Falls: The occurrence of congenital keratoconus posticus circumscriptus in two siblings presenting a previously unrecognized syndrome." Amer. J. Ophthal., 52:53- -57, 1961.

CHIASMATIC SYNDROME

Synonymy

S. of Cushing II.S. of suprasellar meningioma.

Syndromography

Clínic

It usually occurs in adults, at the beginning the patient has a deficit in lateral vision or central vision and later it is characterized by three elements: 1, defect of the bitemporal fields, generally progressive; 2, primary optic atrophy that can lead to total blindness; 3, essentially normal sella turcica.

Paraclinical examinations

Skull radiology. Essentially normal sella turcica, although with modern techniques it is possible to detect some reaction in the sella turcica area. Occasionally, some calcification can be seen in suprasellar meningiomas.

Pneumoencephalogram.

Computed axial tomography and magnetic resonance skull nuclear netics.

Cerebral angiography.

Syndromogenesis and etiology

Suprasellar meningiomas. Other lesions that occupy the region of the optic chiasm, such as craniopharyngiomas, pituitary adenomas, giant aneurysms, chiasmatic gliomas, nasopharyngeal carcinoma, colloid cyst of the third ventricle or remote lesions that produce dilation of the third ventricle and secondarily compression of the optic chiasm.

Literature

Bebin, J. and R.S. Knighton: "Chiasmatic syndrome." Henry Ford Hosp. J., 16:223-233, 1968.

Cushing, H.: "Chiasmal syndrome of primary optic atrophy and bitemporal field defects in adults with normal sella turcica." Arch. Ophthal., 3:505, 1930.

Walker, A.E.: "The neurosurgical evaluation of the chiasmal syndromes." Amer. J. Ophthal., 54:563-581, 1962.

BILATERAL VERTICAL RETRACTION SYNDROME

Syndromography

Clinic

Unilateral or bilateral limitation, total or partial in upward or downward gaze fixation and pseudoptosis. Exotropia

Aya sometimes presents exotropia V. Congenital abnormalities may be associated: unilateral partial ptosis, facial asymmetry and anomalies of the optic nerve.

Paraclinical examinations

Electromyography.

Syndromogenesis and etiology

Hereditary congenital condition.

The etiology is unknown.

Literature

Scassellati-Sforzolini, G.O.: "Una syndrome molto rare: difetto congenito monolaterale della elevation con retrazione del globe." Riv. Otoneurooft., 33:431-439, 1958.

Khodadoutst, A.A. and G.K. von Noorden: "Bilateral vertical retraction syndrome: a family study." Arch. Ophthal., 78:606-612, 1967.

RIEGER SYNDROME

Synonymy

S. of Axenfeld's posterior embryotoxon with juvenile glaucoma. Mesodermal dysgenesis of the cornea and iris. S. from Axenfeld with juvenile glaucoma.

Syndromography

Clinic

Juvenile glaucoma that begins in childhood with great oscillation in tension, frequently associated with neurological manifestations.

Physical examination. Anterior segment disorders: discoria, pseudopolycoria, corectopia, iridostasis, iris dehiscence, prominent iris sphincter, aplasia of the mesodermal leaf of the iris, uveal ectropion, corneal opacification, anterior polar cataract. They are frequently associated with microphthalmos, strabismus, eyelid ptosis and conjunctival heman-gioma.

Associated extraocular manifestations: dental absence or irregularity, hypertelorism, total or partial absence of facial bones.

Paraclinical examinations

Gonioscopy.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. It must be distinguished from

The etiology is unknown.

Literature

Axenfeld, T.: Corneal embryotoxon posterius. Berl. Deutsch. Ophthal. Gesell., 42;AZE, 1920, p. 301.

Montes, J.G. and J.C.G. Montes: "Rieger syndrome: Axenfeld anomaly with familial juvenile glaucoma." Arch. Soc. Oftal. Hisp. Amer., 27:93-99, 1967.

Rieger, H.: "Beitrage zur Kenntnis seitener Missbildungen der Iris; uber Hypoplasie des Irisvorderblattes mit Verlagerung und Entrundung der Pupille." Graefes Arch. Ophth., 133:602-635, 1935.

DERILEY-DAY SYNDROME

Synonymy

S. of family dysautonomia.

Syndromography

Clínic

It usually manifests itself in the first days of life.

Defective tearing, corneal hypoalgesia, relative insensitivity to pain, defects in taste sensation, defects in salivation. Orthostatic hypotension, paroxysmal hypertension, unexplained hyperpyrexia attacks, poor muscle coordination, dysarthria, emotional lability, cyclical vomiting, frequent lung infections.

Physical examination. Frequent corneal ulcers. Absence of tongue papillae, kyphoscoliosis, neuropathic disorders. Areflexia. Romberg sign.

Paraclinical examinations

Characteristic response to certain drugs. Absence of flushing to histamine, exaggerated response to methacholine and norepinephrine, miosis potentiated by the instillation of methacholine into the conjunctival sac.

Radiology of bones and joints.

Electroencephalogram.

Urine. High dosage of vanillyl-manelic acid.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Howard, R.O.: "Familial dysautonomia (Riley-Day syndrome)". Amer J. Ophthal., 64:392-398, 1967.

Riley, C.M.; R.L. Day; D.M. Greeley, et al: "Central autonomic dysfunction with defective lacrimation. I. Report of five cases Pediatrics, 3:468-478, 1949.

ROTH-BIELSCHOWSKY SYNDROME

Synonymy

S. pseudophthalmoplegic.

Syndromography

Clínica

Complete paralysis of all conjugate movements of the eyes in one or more directions, except those movements that are under the control of the labyrinth. Vertical movements are preferentially affected. Stimulation of a maze produces reflex deviation of the eyes to the opposite side of the paralysis; that of the other labyrinth produces deviation of the eyes towards the side of the paralysis.

Syndromogenesis and etiology

Injury to the basal ganglia or the roof of the midbrain.

Bibliography

Bielschowsky, A.: "Das klinische Bild der assozierten Blicklahmung und seine Bedeutung fur die topischen Diagnostik." Munchen. med. Wschr., 50:1666-1670, 1903.

Cogan, D.G and RD. Adams: "Type of paralysis of conjugate gaze (ocular motor apraxia)". Arch. Ophthal., 50:434-442, 1953.
Roth, W.: "Demonstration von kranken mit Ophthalmologie." Neurol. Centralbl., 20:921, 1901.

SCHYNDER SYNDROME

Synonymy

S. of crystalline-corneal dystrophy. S. of hereditary epitheliocorneal dystrophy.

Syndromography

Clinic

It affects both sexes. It begins at an early age in life. Moderate decrease in visual acuity.

Physical examination. The juvenile arch appears early in the form of an opaque, oval or annular cloud in the central part of the cornea with a clear peripheral area. Progressively the opacity extends towards the periphery but never reaches the limbic area.

Paraclinical examinations

Corneal biopsy. It shows normal epithelium and in the opaque area small needle-shaped crystals of unknown composition are observed, located in the anterior portion of the stroma, posterior to Bowman's membrane.

Slit lamp examination.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is unknown.

Literature

Feigin, R.D. and D.B. Caplan: "Corneal opacities in infancy and childhood." J. Pediat., 69:383-392, 1966.
Schynder, W.: "Scheibenformige Kristallenlagerungen in der Hornhautmitte las Erbleiden (hereditary corneal crystalline degeneration)". Klin. Monatsbl. F. Augenh., 103:494-502, 1939.
Van Went, J.M. and F. Wibaut: Hereditary anomaly in cornea". Ned. Tiidschr. Geneesk. 1:2996-2997. 1924.

SCHWARTZ-JAMPEL SYNDROME

Synonymy

S. of congenital blepharophimosis associated with a generalized myopathy syndrome. Diffuse bone disease-dwarfism-myotonia-oculofacial abnormalities.

Syndromography

Clinic

Short stature, hypoplasia of facial bones, small nose. Upward slanted eyes, blepharophimosis, exotropia, myopia, microcornea. Other anomalies: hypertrichosis, arachnodactyly, auricular anomalies, laryngeal hypoplasia, abnormal epiglottis, short neck, pectus carinatus, kyphoscoliosis, acetabulum anomalies.

Paraclinical examinations

Serum enzymes.

Urinary amino acids.

Eromosomic study. Normal pattern.

Radiology. Osteoporosis.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Aberfeld, D.C.; L.P. Hinterbuchner and M. Schneider: "Myotonia, dwarfism, diffuse bone disease and unusual ocular and facial abnormalities (A new syndrome)." Brain, 8:313-322, 1965.
Marden, P.M. and W.A. Walker: “A new generalized connective tissue syndrome; association with multiple congenital anomalies." Amer. J. Dis. Child., 112:225-228, 1966.
Schwartz, O. and R.S. Jampel: "Congenital blepharophimosis associated with a unique generalized myopathy." Arch. Ophth., 68:52-57, 1962.

SIEGRIST SYNDROME

Synonymy

S. of pigmentation of choroidal vessels.

Syndromography

Clinic

It is more common in older women. It is manifested by decreased vision, exophthalmia, granular pigmented areas in the choroid that follows the path of the choroidal vessels. Arterial hypertension.

Paraclinical examinations

Urine. Albuminuria.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Geeraets, WJ.: Ocular Syndromes. Philadelphia, Lea & Febiger, 1965.

Siegrist, A.: Zur Kenntniss der Arteriosclerose der Augengefasse. IX Cong. international dopht. d'Utrecht., 1900. pp. 131-139.

STRING SYNDROME

Syndromography

Clínic

It appears as a complication of the cerclage-type suture used in retinal detachment. It occurs between 4 to 19 days after the operation. It begins with intense pain in the eye, edema of the eyelids, proptosis of the eyeball, chemosis of the conjunctiva, uveitis, ocular hypertension. The cornea remains clear, the anterior chamber is deep and the iris takes on a green color.

Syndromogenesis and etiology

Complication of cerclage surgery. The suture causes vascular obstruction and gives rise to clinical symptoms

The configuration of the eye is a predisposing factor.

Literature

Mason, N.: "The 'string syndrome'; seen as a complication of Arruga's cerclage suture." Brit. M. Ophth., 48:70-74, 1964.

TAPETAL-LIKEREFLEX SYNDROME

Syndromography

Clínica

Ring-shaped scotoma, the retina and choroid have bright yellow-green spots in the region of the posterior pole. Retinitis pigmentosa may be associated.

Syndromogenesis and etiology

Sex-linked heterozygous transmission.

The etiology is unknown.

Literature

Ciccarelli, E.C.: "A new syndrome of tapetal-like fundic reflexes with ring scotomata; report of two cases." Arch. Ophthal., 67:316-320, 1962.

Niccol, W.: "A family with bilateral developmental defects of the macula." Trans. Ophthal. Soc. U.K., 58:763, 1938.

TERSON SYNDROME

Syndromography

Clinic

Equivalent distribution between the two sexes. It predominates in the middle age of life. There are hemorrhages within the vitreous humor, which coexist with all the symptoms and signs of subarachnoid hemorrhage.

Paraclinical examinations

Spinal puncture. Presence of blood.

Angiography.

Syndromogenesis and etiology

The most common cause is the rupture of an aneurysm of an intracranial vessel.

Literature

Castren, J.A.: "Pathogenesis and treatment of Terson syndrome." Acta Ophth., 41:430-434, 1963.

Paton, L.: "Ocular symptoms in subarachnoid hemorrhage." Trans. Ophthal. Soc.U., 44:110-126, 1924.

TOXOPLASMOTIC SYNDROME

Syndromography

Clinic

It occurs in the newborn. It is manifested by strabismus, nystagmus, opacity of the lens, atrophy of the optic nerve, internal hydrocephalus, brain calcification, seizures.

Foci of bilateral chorioretinitis are observed, especially macular, which when fully developed have the appearance of colobomas.

Syndromogenesis and etiology

The syndrome is caused by toxoplasma that is transmitted from the mother to the fetus via the transplacental route.

Literature

Maggiore, L.: Manual of Ophthalmology. Editorial Científico-Médica, Barcelona, 1953. p.691.

POSTERIOR SEGMENT TRAUMATIC SYNDROME

Syndromography

Clinic

It is characterized by retinal, subretinal and periretinal hemorrhages, posthemorrhagic atrophies and pigmentations, choroidal tears, proliferating retinitis, retinal tears and detachments, traumatic macular lesions and ascending atrophies of the optic nerve.

Syndromogenesis and etiology

Trauma that injures the posterior part of the eyeball, especially common when the offending agent acts in the inferoexternal periorbital region.

Literature

Casanovas, José: Ophthalmology. Ed. Rocas, Barcelona, 1961. p. 370.

TÜRK SYNDROME

Syndromography

Clínica

Limitation of abduction of the affected eye beyond the midline, retraction of the abducting bulb.

Syndromogenesis and etiology

The possibility of a birth injury is assessed. It is considered to be an incomplete Duane syndrome, with fibrous degeneration of the lateral rectus muscle.

Literature

Türk, S.: "Ueber Retractionsbewegungen der Augen." Deutsche med. Wschr., 22:199-201, 1896.

UVEITIS-RHEUMATOID ARTHRITIS SYNDROME

Synonymy

S. of Davis.

Syndromography

Clinic

It is more common in children, but can occur at all ages. There are symptoms of rheumatoid arthritis. Visual disorders, pain, tearing and photophobia.

Physical examination. Those described in rheumatoid arthritis.

Uveitis, iridocyclitis; less common are scleritis, adhesion formations in the form of corneal bands and choroiditis.

Paraclinical examinations

Rheumatoid arthritis test. Positive.

Erythrosedimentation. Accelerated.

Syndromogenesis and etiology

Possible autoimmune collagen disease.

The etiology is unknown.

Literature

Davis, M.D.: "Endogenous uveitis in children: associated band-shaped keratopathy and rheumatoid arthritis". Arch. Ophth., 50:443-454, 1953.

UVEOMENINGEAL SYNDROME

Synonymy

Harada disease.

Syndromography

Clinic

It is almost exclusive to the yellow race, it predominates in the male sex, its duration is 2 to 8 months. It is characterized by acute, bilateral exudative choroiditis, with progressive retinal detachment, accompanied by general and nervous alterations (sometimes also of the ear, hair system and skin).

As a consequence of this condition, depigmented areas remain in the choroid.

Paraclinical examinations

Cerebrospinal fluid. Shows lymphocytes.

Literature

Maggiore, L.: Manual of Ophthalmology. Editorial Científico-Médica, Barcelona, 1953. pp.689-690.

UYEMURA SYNDROME

Synonymy

S. of night blindness.

Syndromography

Clinic

It is more common in men, children or young adults. Loss of night vision.

Physical examination. Fund of eye with grayish-white appearance and color and densely covered by yellow-white spots.

rillentas.

Syndromogenesis and etiology

Vitamin A deficiency. A differential diagnosis must be made with punctate retinitis.

Literature

Fuchs, A.: *White spots of the fundus combined with night blindness and xerosis (Uyemura's syndrome)". Amer. J. Ophthal., 48:101-103, 1959.

Uyemura, M.. *Ueber eine merkwurdige Augenhintergrundveranderung bei zwei Fallen von idiopathischer Hemeralopie".

VERTEX SYNDROME

FROM THE ORBIT

Synonymy

S. de Rollet. S. of Rochon-Duvigneaud sensorimotor ophthalmoplegia. S. of painful ophthalmoplegia. S. of the sphenoidal fissure. S. of the sphenoidal fissure-optic canal.

Syndromography

Clinic

Its presentation is sudden, sometimes secondary to respiratory infections. There is decreased vision or total loss, diplopia. Severe pain in the retroorbital and temporoparietal region (ophthalmic area. trigeminal branch).

Physical examination. Slight displacement of the bulb, sometimes absent, from limitation of movement in various directions to complete ophthalmoplegia. Fixed bulb, right forward, due to paralysis of cranial nerves III, IV and VI.

Paraclinical examinations

Skull radiology.

Angiography.

Blood. Leukocytosis and elevated erythrocyte sedimentation rate in some cases.

Cerebrospinal fluid. Variable findings: normal or decreased glucose. Normal or slightly elevated proteins and cells.

Syndromogenesis and etiology

Infections, cysts, aneurysms, neoplasms or trauma to the sinuses (mainly frontal and ethmoidal) that affect the sphenoid fissure. To a lesser extent, it can be caused by non-specific inflammation of the cavernous sinuses.

Literature

Banks, P.: "The superior orbital fissure syndrome." Oral Surg., 24:455-458, 1967.

Hirschfeld, L.: "Epanchemant de sang dans le sinus caverneux du cote gauche diagnostique pendant la vie." Compt. Yield. Soc. Biol., 138. 1858.

Rochon-Duvigneaud, A.: "Quelques cas de paralysie de tous les nerfs orbitaires (opthalmoplegie totale avec amaurose et anesthé sie dans le domaine de lóphthalmidque d'origine syphilitique)". Arch. d'Opth., 16:746-760, 1896.

VOGT-SPIELMEYER SYNDROME

Synonymy

S. of juvenile amaurosis with familial idiocy. Juvenile macular brain dystrophy. S. de Stock-Spielmeyer-Vogt. S. de Spielmeyer-Vogt. S. de Batten-Mayau.

Syndromography

Clínic

It affects both sexes, it is more common in people from Northern Europe or their descendants, but it occurs in any ethnic group.

It begins in the fifth or sixth year of life after a normal childhood. It is characterized by progressive loss of vision, progressive asthenia, apathy, emotional instability, finally seizures and paralysis. Gradual mental deterioration and dementia.

Physical examination. Optic atrophy and pigmentary degeneration of the retina. The typical cherry stain is absent. Ataxia, areflexia. Motor paralysis of the upper and lower neurons.

Paraclinical examinations

Electroencephalography.

Cerebrospinal fluid. Increased proteins.

Other tests. Negatives.

Literature

Jervis, G.A.: “Juvenile amaurotic idiocy.” Amer. J. Dis. Child.. 97:663-667, 1959.

Spielmeyer, W.: "Histologische und histopathologische Arbeiten uber die grosshirnrinde klinische und anatomische Untersuchungen uber eine Besondere Form von familiarer amaurotischen Idiotie." Nissl. Arbeit., 2: 193-213, 1908.

Stengel, C. (1826) reprinted in Nissen, A.J.: "Juvenil amaurotisk idioti i Norge". Nord. Med., 52:1542-1546, 1954.

Vogt, H.: Uber familiare amaurotische idiotie und verwandte Krankheitsbilder". Monatschr. Psychiat. Neurol., 18:161- -171; 310-357, 1906.

VON HERRENSCHWAND SYNDROME

Synonymy

S. of sympathetic heterochromia.

Syndromography

Clinic

Unilateral enophthalmos, heterochromia (unilateral iris).

miosis, facial hypohidrosis on the same side as the lesion.

Syndromogenesis and etiology

Cervical sympathetic paralysis due to: enlarged lymph nodes, tumor, cervical rib or scars.

Literature

Von Herrenschwand, F.: "Ueber verschiedende Arten von Heterochromia iridis. " Klin. Mbl. Augenh., 60:467-494, 1918.

Von Herrenschwand, F.: "Zur Sympathicusheterochromie." Klin. Wschr., 2:1059, 1923.

RELATED OPHTHALMOLOGICAL SYNDROMES

Syndromes with corneal dystrophy (hereditary)

Biber-Haab-Dimmer.

Hereditary deep non-progressive corneal dystrophy.

Hereditary corneal epithelial dystrophy.

Groenouw I.

Groenouw II.

Schnyder.

Orbital fissure syndromes

Superior orbital fissure.

Foix.

Jacob.

Orbit pseudomotor.

Vertex of the orbit.

Oculocerebral syndromes

Aicardi.

Barsy-Moens-Dierckx.

Bloch-Suzlberger.

Cross-McKusick-Breen.

Denys-Corbeel.

Edwards.

François II

Lowe.

McCange.

Patau.

Peters.

Pinsky-di George-Harley-Baird.

Reese.

Smith-Lemly-Opitz.

Sjögren-Larsson.

Ullrich-Feichteiger.

Oculodental syndromes

Dentofacial (Weyers-Fülling).

Dutescu-Grivu-Fleischer-Peters.

Franceschetti.

François II.

Goldenhar.

Hallerman.

Iridodentary (Weyers).

Lejeune.

Lenz.

Oculodentdigital.

Oculootovertebral.

Oculovertebral (Weyers-Thier).

Rubinstein.

Russell.

Rutherfurd.

Ullrich-Feichtiger.

Ullrich-Fremery-Dohna.

Wilderwanck.Marchiafava-Micheli.

Rheumatoid arthritis.

Scleroderma.

Waldenstrom I.

Neurogenic conditions

Carpal tunnel.

Diseases of the nervous system.

Compression of the shoulder ring (girdle).

Post-traumatic conditions

Frosbitc.

Jackhammer.

After the surgical attack.

Vasospasm of pianists and typists.

Occlusive arterial diseases

Arteriosclerosis obliterans.

Embolism.

Thromboangiitis obliterans.

Thrombosis.

Poisoning

Ergotamine.

Heavy metals.

Raynaud.

Syndromes with vascular lesions

Not Fordyce

relatives.

Hasabach-Meritt.

Klippel-Trenaunay-Weber.

Maffucci.

Jejunal-oral-scrotal phlebectasia.

Dominant family member

Blue vesicular-elastic nevus.

Fabry.

Louis-Bars.

Rendu-Osler-Weber.

Sturze-Weber.

Von Hippel-Lindau.

Wyburn-Mason.

Post-heart surgery syndromes

Postcardiotomy diphasic.

Postperfusion pulmonary congestion.

Postcommissurotomy, postcardiotomy.

Postperfusion.

Postpericardiotomy.

Syndromes with syncope

Temporary loss of consciousness observed in different conditions

Adams-Stokes.

Bouveret.

Hypertensive encephalopathy

Cerebrovascular diseases.

Aortic stenosis.

Epileptic.

Heberden.

Idiopathic orthostatic hypertensive.

Pulmonary hypertension.

Hyperventilation.

Hysteria.

Migraine.

Myocardial infarction.

Syncopal due to involuntary urination.

Cough.

Vasovagal.

Weiss-Baker.

Necrotizing vasculitis syndromes

Bazin.

Cutaneous-intestinal-oropharyngeal ulcerations.

Degos.

Acute febrile neutrophilic dermatosis.

Erythema elevatum duitinun.

Erythema nodosum.

Lethal midline granuloma.

Wegener's granulomatosis.

Horton.

Kussmaul-Mayer.

Lindberg.

Löfgren.

Schönlein-Henoch.

Strauss-Churg-Zak.

Cutaneous-intestinal-oropharyngeal ulcerations.

Urbach.

Allergic vasculitis.

Vilanova-Aguade.

Whitfeld.

Zeek.

COSTEN SYNDROME

Synonymy

S. of the temporomandibular joint. S. of painful dysfunction of the tenmporomandibular joint.

Syndromnography

Clínic

This syndrome is characterized by pain of moderate intensity in the frontal, parietal and occipital regions, which reaches the ear, tongue and jaws, caused by prolonged chewing, sometimes also presenting deafness, ringing in the ears and dizziness.

Wear of the temporomandibular joint with narrowing or deformation, as well as sensitivity to palpation, is frequently observed.

It is also accompanied by nystagmus, a burning sensation in the mouth and tongue. Upon examination, vesicular lesions, like herpes, are seen in the external auditory canal.

Paraclinical examinations

Jaw x-ray. It can be useful to check osteoarticular injuries.

Syndromogenesis and etiology

Costen syndrome is related to malocclusion, inappropriate action and abnormal position of the maxillary condyle in the glenoid fossa.

It is due to hyperactivity of the jaw joint, which is followed by a loss of the joint, due to resorption of the menisci, condyles and bone.

The symptoms are due to excessive contraction or spasms of the masseter muscles and irritation of the neighboring nerves: auriculotemporal and chorda tympani.

There are myofascial areas that, through unknown mechanisms, give rise to symptoms, when these areas are stimulated by heat, cold or pressure.

Literature

Costen, J.B.: "Classification and treatment of temporomandibular joint problems." J. Michigan Med. Soc., 55:673-677, 1956.

Freese, A.S.: "Costen's syndrome; a reinterpretation." Arch. Otolaryngol., 70:309-3 14, 1959.

CHARLIN SYNDROME

Synonymy

S. of neuritis of the nasociliary or nasal nerve. S. of the ethmoidal nerve. S. of the ciliary ganglion.

Syndromography

Clínic

It is characterized by unilateral sinusitis, accompanied by ocular symptoms: ulcerative keratitis, iritis and cyclitis; as well as pain at the level of the nasal pyramid and/or the internal corner of the eye.

The ocular symptoms are very striking: epiphora, photophobia.

There is swelling, hyperesthesia and unilateral hypersecretion of the nasal mucosa. This symptomatology returns quickly after cocaineization of the nasal mucosa.

Burning pain occurs in the territory of the nasociliary nerve (internal angle of the orbit and wing of the nose) with intense tearing, redness of the eyelid and episcleral hyperemia sometimes with keratitis and iritis.

Syndromogenesis and etiology

Charlin syndrome is due to a neuritis of the nasociliary nerve, which is a branch of the first branch of the trigeminal nerve and innervates part of the ethmoid bone, anterior nares, eye, and eyelid skin. The ciliary ganglion is also injured.

Its genesis involves the release of histamine substances that modify vascular caliber and tension.

Literature

Bakar, A.B.: Clinical Neurology. Hoeber-Harper, New York, 1962.

Charlin, C.: "The nasal nerve syndrome." Arch. Ophthalm. Hispaner., 31: 339, 1931.

Charlin, C.: "Nasal nerve syndrome." Da Méd.:839-, 1930.

Chernikova, T.V.: Charlin syndrome: case". Sovet. Vestnik. Oftal., 7:398-401, 1935.

Dalessio, D.J.: Wolff's headache and other head pain. Oxford University Press, New York, 1972.

PTERYGOPALATIN FOSSA SYNDROME

Syndromography

Clinic

It is characterized by intense, more or less continuous pain in the upper jaw, which is confused with a toothache, towards the region of the last upper molars, as well as altered sensitivity in the cheek and infraorbital foramen.

Later the pain spreads to the lower jaw. There is ipsilateral deafness and then paralysis of the soft palate and anesthesia and paralysis of the pterygoid muscles are established, which causes deviation of the jaw when opening the mouth.

There is also ipsilateral blindness.

Physical examination. In advanced stages, there is externally visible bulging of the temporal fossa.

Paraclinical examinations

Posterior rhinoscopy. Detects slight bulging of the lateral wall of the pharynx. Unilateral or bilateral enlargement of the retropharyngeal glands. In late stages, washing the oral cavity can cause bleeding.

Syndromogenesis and etiology

Most commonly, this syndrome is due to a metastatic lesion in the pterygopalatine fossa. Less frequently it is due to primary lesions arising from the lateral wall of the back of the nose or the sinus of Morgagni.

Literature

Asherson, N.: Trotter's syndrome and associated injuries." J. Laryng. Otol., 65:349-366, 1951.

GRADENIGO SYNDROME

Synonymy

S. of apical petrositis.

Syndromnography

Clinic

This syndrome consists of the presence of persistent otitis media, weakness of the lateral rectus muscle on the same side causing diplopia and ipsilateral retro-orbital pain.

The patient also presents poor general condition and fever, and complains of anterior temporal headache.

Paraclinical examinations

Radiology. The Town and Stenver technique allows the two sides to be compared and the pneumatization and apical involvement to be estimated.

Polytomography. It defines areas of bone destruction with greater accuracy and one or more propagation tracts through the cells can be outlined.

Syndromogenesis and etiology

This syndrome occurs due to the spread of an infectious process to the overlying dura mater that irritates the abducens nerve (VI) as it passes between the peak of the petrous bone and the sphenoid, through a duct formed by the dura mater (Dorello's duct). This infectious process extends to the trigeminal nerve ganglion (Meckel's ganglion) which is located in Meckel's fossa at the tip of the petrous bone.

The infectious process begins in the petrous portion (petrous bone) of the temporal bone, from acute otitis media and mastoiditis.

Literature

Paparella, M.M. and D.A. Shumrick: Otorhinolaryngology. Havana, Editorial Científico-Técnica, 1984. pp.1472.

MIDLINE LETAL GRANULOMA SYNDROME

Synonymy

S. malignant granuloma type Stewart. S. of midline granuloma. S. of malignant granuloma or gangrenescen.

Syndromography

Clinic

This syndrome affects all ages, most frequently between 30 and 50 years old. Preferably in men of the Caucasian race. In other races it is more common in women.

It begins with a prodromal condition lasting a year or more, consisting of nasal obstruction with serous or serohemorrhagic discharge.

Later, when the disease establishes itself, the secretion becomes purulent, and there are areas of necrotic tissue in the nasal cavity, which extend to involve the entire nose and pharynx and form fistulas that open in the skin, vestibular region of the mouth or the gums. Teeth become loose. The language remains intact. The eyes are affected directly by a granulomatous process or as part of the involvement of the adnexa. In late stages, pain or episodes of needle fever may appear.

Later, difficulty breathing, pain in the paranasal areas, nose and eyes, perforation of the nasal septum and saddle deformity are added. There may be perforations of the soft and hard portions of the palate.

It ends with mutilation and destruction of the affected tissues.

It does not evolve below the neck. Patients may lose vision and experience dysphagia.

Paraclinical examinations

Blood. There is anemia, leukocytosis. Accelerated erythrocyte sedimentation. Hypergammaglobulinemia.

Radiology. Pansinusitis. Bone destruction at sites affected.

Biopsy. Noncaseating granuloma with giant cells or without they.

Syndromogenesis and etiology

It has been attributed to autoimmune phenomena and has been related to Wegener's granulomatosis.

A localized hypersensitivity reaction that causes tissue destruction and mutilation has been proposed. The antigen has not been identified and there are no immunological tests that confirm it. sign.

The etiology is unknown.

Literature

Lehlohr, J. F: Lethal midline granuloma, Clinical aspects". Texas J. Med., 61:188-92, 1965.
McBride, P.: "Case of rapid destruction of the nose and face." J. Laryng., 12:64,1897
Stewart, J.P.: “Progressive lethal granulomatous ulceration of nose.” J. Laryng. & Otol., 48:657-701, 1933.

LERMOYEZ SYNDROME

Synonymy

S. of tinnitus-deafness-vertigo.

Syndromography

Clinic

It occurs in patients between 30-40 years old, unlike Meniére's syndrome, which appears at 50-60 years old.

The sequence of tinnitus and deafness that disappear or decrease after vertigo sets in is another differentiating aspect with Meniére's syndrome, in which vertigo occurs first and is followed by tinnitus and deafness.

In addition, there are allergic manifestations, especially urticaria, that sometimes precede this syndrome.

Syndromogenesis and etiology

Lermoyez syndrome is considered due to vasospasm of the internal auditory artery, probably of allergic origin.

Literature

Eagle, W.W.: "Lermoyez's syndrome an allergic disease." Ann. Otol., 57:464, 1948

Lermoyez, M.: La vertige qui fait understand (angiospasme labyrintique)". Press Med., 27:1-3. 1919.

MENIÉRE'S SYNDROME

Synonymy

Meniere's disease.

Syndromography

Clinic

Symptoms usually begin between the ages of 20 and 60. Although it is generally unilateral, between 20% and 30% both ears are affected. This is more common when the disease occurs at an older age and between 2 and 3 years after the initial symptoms. It is characterized by hearing loss that paradoxically improves after the acute attack. Vertigo, which is the most disabling symptom, decreases as the disease continues its course with destruction of the inner ear.

There are continuous or intermittent, non-pulsatile tinnitus that are not affected by carotid pressure.

Aural fullness is also observed.

Paraclinical examinations

Vestibular tests. There is hypoactivity of the affected labyrinth to caloric stimulation, greater the longer the duration of the disease. Subjectively, caloric irrigation triggers the patient's attack, although the perceived intensity of the response is less than the true attack.

Audiometry. Sensorineural hearing loss is detected for low notes at the beginning of the disease, which can progress to flat or sometimes descending notes later.

Auditory reflex thresholds reveal alterations, as does the binaural loudness balance test (when the process is unilateral).

In half of the patients, temporary improvement in hearing thresholds and discrimination scores is achieved by administering glycerol or furosemide orally.

Electrocochleography. It can be useful to detect cases that could become bilateral.

Skull radiology. Normal.

Syndromogenesis and etiology

There have been attempts to relate this syndrome to allergies, which has not been proven. Some endocrine conditions are considered triggers, such as non-myxedema hypothyroid states, as well as pancreatic insufficiency.

It has also been attributed to hematological pathologies and vascular disorders.

Meniere's syndrome has been associated with familial hyperlipoproteinemia types II, II, or IV, as well as with congenital or acquired syphilis, late or early, due to existing vasculitis.

It may also be present in compression of the superior vestibular nerve by a vascular loop or loops in the internal auditory canal and in consecutive severe sensorineural hearing loss or mumps, meningitis and other unknown factors, and also in several members of a family which suggests a genetic etiology.

The basic alteration is believed to be an abnormality of hydroelectrolyte metabolism within the inner ear with decreased absorption of endolymph by the lymphatic sac, for whatever reason.

The etiology is unknown.

Literature

Paparella, M.M. and D.A. Shumrick: Otorhinolaryngology. Havana, Editorial Científico-Técnica, 1984. pp.1866-1868.

OTOSCLEROTIC INNER EAR SYNDROME

Syndromography

Clinic

The patient experiences episodes of dizziness that last between 20 minutes and 6 hours. The episodes usually consist of vague sensations of floating or fainting, although consciousness is never compromised.

During the episodes, no neurological deficit is observed and they do not leave sequelae.

Paraclinical examinations

Caloric test. When performed on the affected ear it qualitatively reproduces the attack.

Special tests. To detect end-organ disease, they give negative results.

Syndromogenesis and etiology

The etiology is due to otosclerosis.

Literature

Paparella, M.M. and D.A. Shumrick: Otorhinolaryngology. Scientific-Technical Editorial, Havana 1984. p.1874.

DEPENDRED SYNDROME

Synonymy

S. of hereditary goiter with deafness.

Syndromography

Clínic

This syndrome of congenital deafness and goiter is transmitted in an autosomal recessive manner, hearing loss can be detected from birth. It was described by Pendred in 1896.

Both sexes are affected equally. There are various degrees of bilateral deafness, of a perceptual nature and in some cases associated with defects in vestibular function; It is characterized by the loss being greater for high tones than for low tones.

Goiter generally appears at puberty or later but can be present in early childhood and is of variable magnitude; initially it is soft and diffuse with a tendency to become nodular in adult life. Most people

Affected individuals are clinically euthyroid, but hypothyroidism may occur even during childhood.

Physical and mental development are generally normal.

Vestibular dysfunction and slow language development may be observed.

Physical examination. In children there is a diffuse increase in the thyroid, in adults it is more clearly nodular.

Paraclinical examinations

Audiogram. Perceptual hearing loss.

Functional study of the thyroid.

Serum T4. Normal or low.

Serum thyrostimulating hormone. Elevated

Perchlorate test. It is a method described to recognize the type of thyroid defect found in this syndrome.

Radio-iodine uptake test. If it is followed by administration of perchlorate or thiocyanate, it depletes 20-50% of the captured iodine.

The absence of high titers of antithyroid antibodies differentiates this state from chronic lymphocytic thyroiditis.

Caloric test. Hypoactive vestibular labyrinth in most patients.

Syndromogenesis and etiology

Autosomal recessive inheritance. Deafness and goiter are independent expressions of the same genetic defect.

There is a peroxidase or iodine deficiency that prevents the complete organization of iodine.

The genetic defect regulates the activity of enzymes necessary for organization. These peroxidases would play an important role in the initial development of the inner ear.

The possibility of intrauterine hypothyroidism that leads to abnormal development of the auditory apparatus is unlikely due to the greater existence of euthyroid cases.

The possibility of a toxic substance influencing the thyroid and the inner ear has not been demonstrated.

Although the pathophysiology of the hearing defect has not been clarified in many cases, hyperostosis of the promontory and the round window and a Mondini-type defect in the inner ear have been described.

Literature

Fraser, G.R., M.E. Morgans and W.R. Trotter: "The syndrome of sporadic goiter and congenital deafness." Quart. J. Med., 29:279, 1960.

Nilson, L.R.: N. Borgfors; I. Gamstorp et al.: "Non endemic goiter and deafness." Paediat Act. Scand., 53:117, 1964.

Pendred, V.: "Deaf-mutism and goiter." Lancet, 2:532, 1896.

Thorn, A.K. and E.F. Scowen: "Genetic studies of the syndrome of congenital deafness and simple goiter." Ann. Hum. Genet., 27:283-293, 1964.

PIERRE ROBIN SYNDROME

Syndromography

Clínica

This syndrome is characterized by micrognathia, glossoptosis and respiratory obstruction associated with a division of the soft palate.

The child presents a typical appearance with a small, retracted chin and with lower alveoli located one or more centimeters behind the upper alveoli.

Respiratory obstruction is due to valve closure of the pharynx caused by glossoptosis. The obstruction can be

It is not very pronounced at the time of birth, but usually increases during the following days as the child becomes weaker due to feeding difficulties. Obstruction is frequently episodic and is accompanied by attacks of cyanosis, stridor, and deep inspiration when the child is in the supine position during feeding or sleeping. Airway patency can be improved by placing the child in a prone position.

Feeding problems are very pronounced due to glossoptosis, which impairs swallowing, so children only ingest small amounts of food and have frequent bronchial aspirations.

The lack of growth and repeated infections caused by bronchial aspiration lead to a worsening of obstructive symptoms and lead to respiratory cachexia syndrome.

Syndromogenesis and etiology

Although in some cases it is of familial origin, the etiology is unknown.

Literature

Deconomopoulas, C.T.: "The Value of Glossopexy Robin the (Pierre) Syndrome." New English J. Med., 262:1267, 1960.

Handley, R.C. and J.B. Johnson: "Utilization of the Kirschner Wire in (Pierre) Robin Syndrome." Plastic & Reconst. Surg., 31:587, 1963.

Routledge, R.T.: "The (Pierre) Robin Syndrome: A Surgical Emergency in the Neonatal Period'." Brit. J. Plast. Surg., 13:204, 1960.

FIRST NEURON SYNDROME

Syndromography

Clinic

It is characterized by perceptual hearing loss; algiacusis is not detected.

Paraclinical examinations

Supraliminal tests. It is found: negative recruitment, normal ceiling, normal differential threshold of intensity and frequency, normal fatigue, negative diploacusis.

Acuphenometry. It shows intense high-pitched tinnitus and it takes 30 decibels or more to deafen the patient. Sometimes it is impossible to bring it to that state despite increasing the intensity.

Logoaudiometric curve. It gradually separates from the normal towards the top and does not reach 100% uptake with any intensity. This is due to the indiscrimination caused by poor or no hearing of high-pitched letters; However, at the highest intensities, since there is no algiacusis, the curve does not go down again, which serves to differentiate this syndrome from cortypathy.

Vestibular involvement, however, is more common in this syndrome than in cortipathies, which is due to the intense relationship that the two roots of pair VII have in their path. This motivates the adoption of the type of harmonic syndrome with vestibular hypoexcitability on the diseased side.

Syndromogenesis and etiology

First neuron syndrome is due to lesions that occur between the ganglion and the nuclei of actual origin of the branch.

cochlear, among which are tumors of the cerebellopontine angle, arachnoiditis of the posterior fossa, meningoneuritis, gummas and exostoses of the internal auditory canal, as well as certain head traumas.

Literature

Adams, Raymond D. and M. Victor: Principles of Neurology. McGraw-Hill, New York. 1985.

PSYCHOPHONASTHENIA SYNDROME

Syndromography

Clínic

This syndrome frequently begins in late adolescence or after stress, professional or personal. It usually occurs in cultured and intelligent people, with a strong personality, but at the same time they are shy and supersensitive.

It is manifested by marked sensitivity in the throat, a feeling of suffocation when they begin to speak, head and neck paresthesia, dryness or excessive secretion of saliva in the mouth. Cracking of the voice and pain in the vocal cords when speaking for a long time.

Occasionally it is accompanied by tachycardia, palpitations, and sweating.

Syndromogenesis and etiology

This syndrome is considered to be a neurotic manifestation, derived from a feeling of imperfection.

Literature

Greene, J.S.: “Psychophonasthenia syndrome.” Ann. Otol. Rhine. Laryng., 50:177-84, 1941.

SLUDER SYNDROME

Synonymy

S. of lower facial neuralgia. S. of the sphenopalatine ganglion. S. of neuralgia of the vidian nerve.

Syndromography

Clinic

It is characterized by pain that begins in the roof of the nasal passages around the eyes, in the upper jaw and teeth, and radiates backwards to the temporal region and the ear, as well as to the back of the neck and even to the shoulder.

Its occipital irradiation differentiates it from trigeminal neuralgia. Painful crises are recurrent and last minutes, hours or days.

The pain may become more intense at night but its radiation is not linked to the course of the three branches of the vidian nerve.

Vasomotor rhinitis, tear flow, and conjunctival injection are frequently associated. Nasal symptoms predominate in this syndrome.

Syndromogenesis and etiology

It is due to neuralgia of the vidian nerve, caused by irritation in the mucosa of the nasal passages and adjacent sinuses.

tes, especially the maxillary sinus, posterior portion of the ethmoid and sphenoid sinus.

Literature

Caviness, V.S. and P. 0'Brien: "Headache", New. Engl. J. Med.. 302:406, 180.

Diamons, S.: "Prolonged benign exertional headache." Headache, 22:96, 1982.

Sluder, G: "The role of splenopalatine (Meckel's) ganglion in nasal headaches." New York Med. J., 87:989-990, 1908.

USHER SYNDROME

Synonymy

Retinitis pigmentosa and sensorineural deafness. S. of retinitis pigmentosa-deafness.

Syndromography

Clinic

Affected patients do not present symptoms until they are 6 years old, when deafness begins. Visual defects begin a few years later due to night blindness and reduction of the visual field. Visual symptoms are what determine medical attention and deafness is detected during paraclinical examinations.

Eye examination demonstrates pigmented retina and cataracts may be present.

Some patients present vestibular symptoms and loss of olfaction. Aphasia and mental retardation at times.

Clinical variations have determined that this syndrome is classified into four types:

Type I. Profound deafness with onset of retinitis pigmentosa (R.P.) at 10 years of age.

Type II. Severe to moderate deafness with R.P. onset after 10 years.

Type III. Diagnosis of R.P. before deafness which is progressive.

Type IV. Possible X-linked recessive form.

Most cases are type I.

Paraclinical examinations

Audiometry. Diagnosis of deafness.

Campimetry. Visual field defect.

Dark adaptation. Flaw.

Electroretinogram. Abnormal.

Syndromogenesis and etiology

It is postulated that hearing loss and retinitis pigmentosa represent a disorder of the central nervous system that produces pathological changes in the brain and also degeneration of neuroectodermal tissues in the terminal organ.

The etiology is genetic with genetic heterogeneity. There may be a relationship between the genetic defect and the degenerative effects of the specialized cells of the inner ear and the photoreceptors. The basic defect is unknown.

Literature

Davenport, S.L. et al.: "Usher syndrome in four hard-of-hearing sibbling." Pediatrics, 62:578-583, 1978.

Paparella, M.M. and D.A. Shumrick: Otorhinolaryngology. Havana, Editorial Científico-Técnica, Havana, 1984. pp:1713-1714.

RELATED ENT SYNDROMES

Syndromes with hereditary deafness

Hereditary deafness with ectodermal alterations

Klein-Waardenburg.

Tietz.

Hereditary deafness with ear malformations

External.

Escher-Hirt.

Fouman-Fouman.

Fárá-Klup ckov -Hrivnakova.

Mengel-Konigsmarck-Berlin-McKusik.

Rowley.

Wilderwanck.

PARANEOPLASTIC SYNDROMES

INTRODUCTION

In recent years we have witnessed notable progress in the knowledge of the biological and biochemical bases of cancer, which does not mean that the problem of neoplastic diseases has been solved. Advances in oncological treatment of adults have been gradual and have focused on malignant diseases characterized by extraordinary sensitivity to radiation and chemotherapy; These, among others, are acute myelocytic leukemia, lymphoproliferative malignancies, testicular cancer and breast cancer. New treatment modalities such as immunotherapy and agents that promote normal cell maturation are under intensive and experimental research. Meanwhile, research has been initiated in search of compounds that can interact with the products of oncogens, genetic regulators, growth factors, and their receptors, etc.

Modern technology in molecular genetics and immunology may have, in the near future, new anticancer drugs.

The terms cancer, neoplasia and malignant disease are often used interchangeably in both technical and popular literature. The disease called cancer is best defined by four characteristics that describe the different way cancer cells act compared to the way normal cells act:

1 Clonality. In most cases, cancer originates from a "parent" cell that proliferates to form a clone of malignant cells.

2. Autonomy. Growth is not regulated appropriately by the normal biochemical and physical influences in the environment.

3. Anaplasia. There is a lack of coordinated normal cellular differentiation. The cells tend to have large, deformed nuclei with more notable chromatin and prominent nucleoli; increased mitosis and abnormalities in some cells and/or giant cells with multiple nucleoli, reflecting aneuploidy, failure of karyokinesis, or both. When abnormal cells are found but do not meet the criteria for anaplasia (loss of differentiation), they are called dysplastic. The dysplasia observed in premalignant lesions may be reversible.

4. Metastasis. Cancer cells develop the ability to grow discontinuously and spread to other parts of the body. This is because they lose their adherence and fixed position within an organized tissue, move to adjacent sites, develop the ability to invade and exit blood vessels, and become capable of proliferating in unnatural sites or environments.

Cancers produce clinical symptoms in three general ways:

1. Due to direct effects due to invasion or compression of normal tissues.
2. By release of cytokines, hormones and other biologically active agents, to the local or general environment.
3. Due to secondary psychological effects on the patient.

Paraneoplastic syndromes include symptoms produced by the release of biologically active agents, as well as systemic problems due to mechanisms not yet determined. Generally speaking, in these syndromes, severe functional effects in the patient may be caused by excessive synthesis of a product of a gene that is normally in that particular cell type, or the malignant cell may produce a molecule that does not exist in the similar normal cell.

In principle, the symptoms that constitute paraneoplastic syndromes have no anatomical relationship with the neoplasia or its metastases, although they evolve in parallel with it. The term was created in 1961 by Boudin and includes the syndromes, symptoms or biological alterations that evolve before, during or after a neoplasia, without having a direct relationship with it or its metastases as already expressed.

The reason for including this small chapter in the book is due to the great usefulness that its knowledge can provide to the doctor, to establish the early diagnosis of cancers. It should be noted that if the original neoplasia is diagnosed and treated early, the clinical picture of the present paraneoplastic syndrome will also disappear.

The syndrome may accompany or mask the primary tumor and evoke a second condition that may even be diagnosed biologically or histologically, whose symptoms may be more intense and not correlate with the original tumor.

Currently, more than 70 paraneoplastic syndromes have been described in the literature and their number is increasing as human body examination techniques develop.

These syndromes remain generally poorly known (except for some more classic ones) and their epidemiology has been little studied; However, it is estimated that around 15% of neoplasms present symptoms compatible with a paraneoplastic syndrome.

It would be good to remember that from the moment Olivier d' Angers described in 1837, the destructive attack of the meninges and spinal roots by a metastatic carcinomatous process, until the demonstration of a paraneoplastic degeneration of the posterior cords of the medula described in 1948 by Denny Brown, 111 years passed. This relatively long interval constitutes a good testimony of the doctor's interest and hesitations to understand and, above all, to elucidate numerous aspects and clinical translations of these syndromes.

We will review and briefly point out the main paraneoplastic syndromes grouped by system.

Some will be described in more detail to make it easier for readers to understand.

DERMATOLOGICAL PARANEOPLASTIC SYNDROMES

Skin manifestations of non-metastatic origin precede or follow cancer in fairly short periods and their evolution is parallel. Sometimes they come from or occur after the diagnosis of certain tumors.

They can be classified into:

1. Malignant skin neoplasms that imply a high risk of an independent primary malignant process:

a) Bowen syndrome.

b) Kaposi's sarcoma.

2. Skin changes due to tumor metabolic products:

a) Malignant carcinoid syndrome. Episodic redness of the face and neck produced by serotonin and other vasoactive peptides (medullary thyroid carcinoma and carcinoids). It is worth remembering that alcohol, glutamate and certain drugs, such as chlorpropamide, calcium channel blockers and nicotinic acid, can produce flush reactions similar to carcinoid syndrome.

b) Addisonian hyperpigmentation with Cushing syndrome, from carcinomas that produce MSH or ACTH-like peptides.

c) Generalized dermal melanosis, slate gray (malignant melanoma).

d) Nodular fatty necrosis due to lipases formed from pancreatic carcinoma.

e) Raynaud's phenomenon with cryoproteinemia (multiple myeloma).

f) Amyloidosis (multiple myeloma, 10%).

g) Necrolytic migratory erythema (glucagonoma). Area of vesicular and erosive erythema that affects periorificial and intertriginous areas. Frequent stomatitis.

h) Porphyria cutanea Tarda secondary to primary hepatoma.

river.

i) Eosinophilic arthralgic panniculitis (pancreatic tumor).

3. Skin changes due to disorders in the function of other systems induced by non-endocrine malignant processes:

a) obstructive jaundice.

b) Addisonian hyperpigmentation due to tumor infiltration of the adrenals.

c) Thrombocytopenic purpura.

d) Paleness due to anemia.

e) Herpes zoste.

f) Severe, prolonged, recurrent herpes simplex.

g) Recurrent pyoderma.

h) Very intense delayed hypersensitivity due to mosquito bites.

4. Idiopathic skin changes:

a) Changes often related to internal malignant processes:

- Dermatomyositis in adults (genital carcinoma

- Acanthosis nigricans, hyperkeratosis and hyperpigmentation in the armpits, neck, flexures and anogenital region produced by IGF-alpha.
  Gastric cancer 60%, sometimes squamous or undifferentiated cell carcinoma or lymphomas.

- Migratory thrombophlebitis (pancreatic carcinoma).
  Scleroderma (visceral carcinoma).

- Ichthyosis in adults (lymphoma, most common Hodgkin's).

- Mucinous alopecia in adults (lymphoma).

- Acquired pachydermoperiostosis (bronchial carcinoma, also in: lung infections, congenital heart disease or liver disease).

-Acquired villous hypertrichosis (*malignant lanugo"), Herzberg-Potjan-Gebauer syndrome (carcinomas of the breast, bladder, lung, gallbladder, colon or rectum).
Erythema gyratum repens, Gammel syndrome: urticarial manifestations arranged in the form of concentric arched lesions that resemble the grain of a wood or the image of a zebra on the trunk, with scabs and itching (bronchial and breast carcinomas).

b) Changes occasionally related to internal malignant processes:

- Pruritus without causal skin lesions (Hodgkin's, 30%; mycosis fungoides, almost universal).
  Clubbing fingers, possibly with hypertrophic osteoarthropathy.

- Erythroderma (lymphoma, leukemia and reticulosis).

- Normolipemic xanthomatosis (neoplasms of the reticuloendothelial system, 50%, multiple myeloma).

- Erythema multiforme (lymphoma, leukemia, carcinoma).

-Urticaria pigmentosa (Nettelship syndrome) and erythema perstans: persistent papular lesions that change shape (concentric, annular, polycyclic).

- Atypical pyoderma gangrenosum, febrile neutrophilic dermatosis (myelogenous and myeloblastic leukemia, myeloid metaplasia, monoclonal gammopathy and polycythemia).

-Vesicular disease (vesicular pemphigoid and Duhring's dermatitis herpetiformis).

-Sudden onset multiple seborrheic keratosis (Lesser-Trelat sign). Produced by TGF-alpha, it is observed in digestive adenocarcinomas and lymphomas.

5. Hereditary diseases with skin manifestations and propensity to develop internal malignant neoplasia:

a) Autosomal dominant inheritance:

- Multiple hamartoma syndrome (Cowden's).
  Thyroid and breast carcinomas.

- Gardner syndrome. Colon adenocarcinoma (very high frequency).
  Multiple mucosal neuromas. Pheochromocytomas, medullary thyroid carcinoma (high frequency).
  Neurofibromatosis (Recklinghausen's). Malignant neurilemmoma (frequency S5%), pheochromocytoma (rare), astrocytoma, glioma (rare).
  Nevoid basal cell carcinoma syndrome.
  Medulloblastoma, fibrosarcoma of the jaw (low frequency).
  Palmo-plantar hyperkeratosis (tylosis, Clarke syndrome).
  Esophageal carcinoma (frequency 95%).
  Peutz-Jeghers syndrome. Gastric, duodenal and colon adenocarcinomas (low frequency).

- Tuberous sclerosis. Astrocytoma, glioblastoma (low frequency).

b) Autosomal recessive inheritance:

- Ataxia-telangiectasia. Lymphoma, leukemia (frequency 10%).

- Bloom syndrome. Leukemia (high frequency).

Chediak-Higashi syndrome. Lymphoma (high frequency).

- Fanconi anemia. Leukemia (high frequency).

- Werner syndrome (adult progeria). Sarcoma, meningioma (frequency 10%).

c) Sex-linked recessive inheritance:

Sex-linked Bruton's aganmaglobulinemia.

Leukemia, lymphoma (frequency 5%).

Dyskeratosis congenita. Carcinomas (high frequency).

Leukemia (occasional).

- Wiscott-Aldrich syndrome. Leukemia, lymphoma (frequency 10%).

NEUROLOGICAL PARANEOPLASTIC SYNDROMES

In 1965, Croft and Wilkinson estimated that 7% of patients with tumors had neuromuscular disorders that could not be explained by local tumor proliferation. They constitute a series of specific syndromes that can affect the central nervous system or the peripheral nervous system, the neuromuscular junction, or the muscle individually or together, which occur in patients with cancer, sometimes even preceding their diagnosis. There is increasing evidence that some of them are related to the presence of autoantibodies or antibodies initially directed against the neoplasia, and that cross-react against certain neural groups (Purkinje cells, dorsal ganglion neurons).

They appear in 1% of cancer patients. Its diagnosis is important because eventually the paraneoplastic syndrome is an early manifestation and guides towards the diagnosis of cancer. Other times it should not be confused with a compressive neurological phenomenon and sometimes the initial evolution of the neurological disorder can be more serious than that of the tumor itself.

In recent years, it is considered that the etiopathogenesis of neurological paraneoplastic syndromes is fundamentally due to a viral mechanism originating from an opportunistic infection with viruses from the papova group, as appears to occur in progressive multifocal leukoencephalopathy and subacute motor neuropathy.

Another important possibility is that of an autoimmune mechanism, with circulating antibodies found against Purkinje cells, neuronal nucleus (Anti-Hu), anti-cytoplasm of Purkinje cells and anti-cytoplasm of retinal ganglion cells.

A classification grouped according to location and/or manifestation clinical presentations in relation to the neuromuscular system is more practical:

1. Brain:

a) Limbic encephalitis (oat cell lung carcinoma).

b) Degeneration of photoreceptors (oat cell lung carcinoma).

) Progressive multifocal leukoencephalopathy (bronchial carcinoma, Hodgkin, lymphomas, leukosis; also associated with: tuberculosis, sarcoidosis and papovavirus).

d) Encephalopathies of biologically active endocrine tumors.

e) Paraproteinemia encephalopathies (Bing-Nel syndrome)

2. Cerebellum and brain stem:

a) Subacute degeneration of the cerebellar cortex (carci-oat cell lung noma, breast cancer and ovarian cancer, Hodgkin's disease).

b) Opsoclonus-myoclonus (neuroblastoma).

c) Bulbar encephalitis (lung cancer).

3. Spinal cord:

a) Subacute necrotizing myelopathy (oat cell lung carcinoma, lymphomas).

b) Subacute motor neuropathy (non-Hodgkin lymphoma).

c) Amyotrophic lateral sclerosis.

d) Chronic myelopathy (spino-cerebellar degeneration).

4. Roots and peripheral nerves:

a) Subacute sensory neuropathy. Denny-Brown syndrome (oat cell lung carcinoma, other lung cancers). It is characterized by greater impairment of deep sensitivity that can lead to severe ataxia and areflexia, but maintaining strength.

It has been related to the presence of specific antibodies known as anti-Hu. The cerebrospinal fluid is usually normal.

b) Acute polyneuritis. Guillain-Barré syndrome (Hodgkin's disease).
Motor-sensory neuropathy. Wyburn-Masson syndrome (oat cell lung carcinoma).
Two syndromes have also been described: distal sensory-motor neuropathy in Waldeström macroglobulinemia and peripheral neuropathy in osteosclerotic myeloma.

d) Peripheral neuropathy accompanied by monoclonal gammopathy (proliferative disorders of plasma cells and B lymphocytes).

e) Neuropathy with insulinoma (insulinoma).

5. Neuromuscular junction:

a) Myasthenia gravis (thymoma, stomach cancer, breast cancer).

b) Lambert-Eaton syndrome (oat cell lung carcinoma).

6. Muscles:

a) Polymyositis (lung, ovarian, breast cancer, lymphomas) .

Some central neurological syndromes are described in more detail below.

ENCEPHALOMYELITIS SYNDROME

It is characterized by perivascular inflammatory infiltrates composed of lymphocytes. The clinical manifestations correspond to three syndromes:

a) Acute sensory neuropathy. It presents with radicular pain and sensory involvement, especially arthrokinetic and vibratory, beginning in the arms.

asymmetrical, abolition of osteotendinous reflexes with pseudotetotic movements and sometimes involvement of the trigeminal nerve.

b) Involvement of the brain stem and/or the motor neuron of the spinal cord. It presents with manifestations due to alteration of the brain stem, with vertigo, instability, diplopia and alterations in gaze or paralysis of the bulbar cranial nerves with generalized weakness due to involvement of the spinal motor neuron. The condition may suggest Guillain-Barré syndrome.

c) Limbic encephalitis. It is characterized by psychiatric disorders, such as anxiety, depression, hallucinations and psychomotor agitation, memory impairment and impairment of cognitive functions. Later there may be alterations of the brain stem or posterior spinal ganglion. The condition has a subacute onset and the patient may be diagnosed with a psychiatric illness.

CEREBELLAR DEGENERATION SYNDROME

Its incidence is very low, less than 1% and it usually precedes the diagnosis of the tumor. The lesion is characterized by a loss of Purkinje cells in the cerebellum and sometimes degeneration of the posterior cords and spinocerebellar tracts. The clinical picture is subacute with ataxic manifestations, gait alterations and later incoordination of all limbs. Sometimes these manifestations are accompanied by alterations of oculomotor nerves, diplopia, vertigo and nausea. On examination there is intention tremor, hypotonia, incoordination, nystagmus, dysarthria and alterations of the oculomotor nerves.

DEOPSOCLOUS SYNDROME

It is an ocular motility disorder characterized by rapid, irregular, arrhythmic and multidirectional movements sometimes accompanied by myoclonic contractions and acute cerebellar ataxia. The lesion may be due to a lesion of inhibitory neurons that control neuronal groups located in the pons segment and that generate these eye movements.

RETINAL DEGENERATION SYNDROME

It is characterized by acute loss of vision, a consequence of demyelination and axonal loss of the optic nerve. It is not due to direct invasion of the tumor and appears preferably before its diagnosis, in isolation or during the course of paraneoplastic encephalomyelitis. The fundus may be normal.

NECROTIC MYELOPATHY SYNDROME

It is a rare condition characterized by flaccid parapesia, sphincter involvement, and extensive sensitivity disorder.

MOTONEURONE INJURY SYNDROME

Injury to the anterior horn of the cord as a paraneoplastic syndrome includes the following syndromes:

a) Association with amyotrophic lateral sclerosis.

b) Motor neuron injury in paraneoplastic encephalomyelitis.

c) Hemopathies with monoclonal band and motor injury neuron.

d) Subacute neuropathy syndrome associated with lymphoma.

The syndromes that mainly affect the peripheral nerves are:

1. Asymmetrical sensory peripheral neuropathy.
2. Sensory-motor neuropathy.
3. Pure sensory neuropathy associated with degeneration of the posterior root ganglia.
4. Acute ascending polyneuropathy.
5. Vegetative neuropathy.
6. Multineuritis.

These syndromes are characterized by weakness and loss of distal sensitivity with reduction or abolition of tendon reflexes, generally in the lower limbs.

In autonomic neuropathy, ostostatic hypotension appears and manifestations of neurogenic bladder, alterations in esophageal and intestinal peristalsis, and symptoms of intestinal pseudo-obstruction are detected. In the cerebrospinal fluid there is an increase in proteins and electromyography and biopsy demonstrate demyelination and axonal degeneration.

LAMBERT-EATON MYASTHENIC SYNDROME

It is a rare syndrome that appears more frequently in men and is characterized by weakness in the trunk and proximal muscles of the arms and legs. It is associated with dry mouth, dysarthria, dysphagia, blurred vision, diplopia and ptosis. In contrast to myasthenia gravis, muscle strength improves with exercise and there is little response to tension. Tendon reflexes are reduced or absent. The electromyogram (EMG) reveals a low amplitude of muscle action potentials with a single stimulation, followed by a progressive increase in the size of the muscle action potentials with repeated stimulation of the nerve.

DERMATOMYOSITIS AND POLYMYOSITIS SYNDROME

The presence of cancer appears in between 7 and 34% of patients with dermatomyositis or polymyositis. The clinical picture is characterized by muscle weakness that affects the upper muscles. Muscle enzymes are elevated, EMG is abnormal, and the biopsy shows fibrillar necrosis with minimal inflammatory changes.

NEUROPATHY AND HIV INFECTION SYNDROME

The involvement of the nervous system is one of the most frequent and important. Sometimes the data of paraneoplastic syndrome involvement are masked by the coexistence of lesions of the central nervous system. Various clinical forms of neuropathy have been described in relation to infection by the human immunodeficiency virus (HIV), some authors postulating that direct invasion of the nerve by the virus would be the cause.

Neuropathies associated with HIV infection are:

1. Distal symmetrical polyneuropathy:

a) In relation to vitamin B12 deficiency.

b) In relation to neurotoxic drugs.

c) Idiopathic.

2. Inflammatory demyelinating polyneuropathy.
3. Mononeuropathy multiplex.
4. Progressive polyradiculopathy.
5. Autonomic neuropathy.

Distal polyneuropathy

It is the most common form, since in fact it can be detected in the necropsies of almost all patients. When other causes such as vitamin B12 deficiency, alcoholism, diabetes or exposure to neurotoxic drugs are ruled out, 30% of cases remain that are considered idiopathic. They usually present a constitutional syndrome associated with significant weight loss and distal paresthesias without weakness. There may be nonspecific alterations of the cerebrospinal fluid. The conduction speed is slowed, and sensitive action potentials may not be registered.

Inflammatory demyelinating neuropathy

They are relatively acute forms (Guillain-Barré Syndrome) and chronic. Guillain-Barré syndrome can occur in seroconversion. Both forms are more common in asymptomatic seropositive people, although they can also be seen in immunodeficient patients. From a clinical and electrophysiological point of view, they are indistinguishable from the idiopathic forms described in previous sections.

The presence of elevated protein in the cerebrospinal fluid (50-250 mg%) with pleocytosis (20-50% lymphocytes) should raise suspicion of HIV infection. Chronic forms improve with steroids and plasmapheresis.

Mononeuropathy multiplex

Described by Lipkin in 1985, it is characterized by asymmetric multifocal involvement. A limited form with acute involvement of 162 nerves has been described, which may have a favorable spontaneous evolution. There is also an extensive form with 3 or more injured nerves, and it is likely, if these patients present associated immunodeficiency, that it is related to neuropathy with cytomegalovirus (CMV) infection. The changes in the cerebrospinal fluid are nonspecific and neurophysiological studies show axonal damage. Various pathogenic mechanisms can intervene in its development (immune response, cytomegalovirus infection, necrotizing arteritis).

In severe cases of neuropathies with systemic HIV infection, treatment with ganciclovir is indicated.

Progressive polyradiculopathy

Described by Eidelbreg in 1986, it occurs in advanced stages of the disease, coinciding in a large proportion of cases with systemic cytomegalovirus infection. It manifests itself as paresthesias in the lower limbs and sacrum, ascending hypoesthesia, sometimes with a sensory level, paraparesis, areflexia and sphincter alterations. In advanced stages, the upper limbs can be affected. This process is due to a direct CMV infection that has been isolated in cerebrospinal fluid and identified in affected nerves. The response to treatment with ganciclovir is more effective the sooner it is established.

Entity	Affected area	Cancer
Central nervous system:		
Encephalomyelitis!	Hippocampus brainstem Marrow Posterior spinal ganglion	Lung'
Subacute cerebellar degeneration	Purkinje cells	Ovary, breast, lung, Hodgkin lung²
Opsoclonus		Lung' Neuroblastoma
Retinopathy	Ganglion chain	Lung
Necrotic myelopathy	Marrow	Lung
Motor neuropathy	Motoneuron	Lymphoma
Peripheral Nervous System:		
Sensorymotor neuropathy Pure sensory polyneuritis Acute polyneuritis	Axon Posterior ganglia Myelin	Lung, myeloma Lung Hodgking, lung
Vegetative neuropathy	Vegetative ganglion	Lung'
Intestinal pseudo-obstruction	Myenteric plexus	Lung'
Multineuritis	Microvasculitis	prostate, lung
Neuromyopathy	Terminal axon?	Lung
Presynapse and muscle:		
Eaton-Lambert	Muscle	Lung
Dermatomyositis	Presynapse	Lung, breast Lung
Necrotic myopathy	Muscle	

I Small cell lung cancer.
2 Includes subacute sensory neuronopathy.

There are data that confirm the presence of subclinical autonomic neuropathy in HIV-infected patients, with its manifestations (orthostatic hypotension, syncope, impotence, diarrhea) being more frequent in later stages. On the other hand, although patients with motor neuron disease and sensory neuronopathy have been described, their relationship with virus infection is less defined.

The syndromes that mainly affect the central nervous system and the peripheral nerves, and the neoplasms most frequently affect them, are indicated in the following table.

RENAL PARANEOPLASTIC SYNDROMES

Among the possible consequences of neoplasms, the following paraneoplastic manifestations with renal expression are admitted:

1. Nephrotic syndrome (11% associated with cancer). Bronchial, colon, ovarian and uterine carcinomas, otorhinolaryngological tumors and some hemopathies (Hodgkin's, leukosis, lymphomas). In the latter case, the association of malignant hemopathy and nephrotic syndrome without tumor infiltration of the parenchyma remains highly debated. However, proteinuria disappears with tumor excision.
   Ectopic secretion of polypeptide hormones by the tumor: These substances can have direct or indirect consequences on the kidney through electrolyte or water modifications: hypercorticism, which causes hypokalemia, alkalosis; hypersecretion of ADH, which causes edema and hemangioblastoma (oliguria); hypersecretion of parathyroid hormone, which causes hypercalcemia.
3. Tubular syndromes. They are rare and are due to tubular obstruction by cells in cases of myeloma (Fanconi syndrome).

PARANEOPLASTIC SYNDROMES

OSTEOARTICULARS

From a clinical point of view, painful osteoarticular symptoms have a pseudorheumatic appearance and biologically resemble seronegative atypical polyarthritis, and evolve in parallel with the tumor signs. This cancer-rheumatism relationship has been known for a long time and can be associated with multiple forms of tumors, particularly pleural mesothelioma for example, which produces intractable arthralgia in more than 60% of cases.

The most typical syndrome continues to be the hypertrophic pneumatic osteoarthropathy of Pierre-Marie-Strumpell-Bamberger, which is characterized by dysachromelic alterations, to which it is associated: gynecomastia, vasomotor disorders and osteoarticular pain. In 85% of cases it accompanies squamous cell carcinoma of the lung and the rest of the cases are divided among non-tumor conditions: bronchopulmonary suppurations, hydatid cyst, ulcerative colitis, liver cirrhosis, cyanogenous heart disease and finally other extrathoracic tumors.

Rheumatic polymnialgia is often associated with Hodgkin's disease and also with esophageal cancer.

The inflammatory syndrome (metacarpophalangeal and interphalangeal joints, elbows and wrists) is associated with carcinoid syndrome.

It is good to specify that carcinoid syndrome, caused by carcinoid tumors, tends to be considered related to apudomas, according to Pearse's concept of APUD tumors (A: strong concentration of amino acids: PU: which have the capacity to produce amine precursors; D: presence of decarboxylases). These tumors arise from the chromoargentafine cells of the digestive mucosa, but also from the bronchial and ovary. It is proposed that the production of hormones in carcinoid syndrome originates in the Kulchintsky cells in which the production of bombesin (gastrin-releasing peptide) has been demonstrated. Carcinoid syndrome is characterized by intense secretory activity of:
serotonin (5-hydroxyindoleacetic acid or 5-hydroxytryptamine), bradykinin, histamine, prostaglandins, ACTH and sometimes thyrocalcitonin. 30% of these tumors are located in the small intestine (most frequent clinical manifestations) and to a lesser extent in the bronchi (bronchial carcinoma).

PARANEOPLASTIC SYNDROMES WITH METABOLIC DISORDERS

Hypercalcemia syndrome

Already in 1885, Virchow described hypercalcemia as a complication of malignancy. Clinical symptoms can be seen in this syndrome. Hypercalcemia is associated with: kidney neoplasms (30%), bronchial cancer (20%), the rest among urogenital, breast and digestive cancers. It is good to remember that 6% of all neoplasms present hypercalcemia without signs of metastasis.

Hypoglycemia syndrome

Extrapancreatic tumors causing hypoglycemia are rare and concomitant insulin formation is the rule.
Until 1981, 180 cases had been published. A histological classification groups them as follows: 1, mesenchymal tumors; 2, epithelial tumors (liver, gastrointestinal, adrenal, breast and lung).

In relation to mesenguimal tumors, until 1981, 84 cases had been published. These were fibrosarcomas, neurofibromas, lymphosarcomas and multiple cases of lipoleiomyorabdosarcomas, as well as mesotheliomas and hemangiopericytomas. Their location is fundamentally retroperitoneal or sometimes intrathoracic; When associated with goiter and hyperthyroidism, they are known as Doege-Potter syndrome.

The best-known epithelial tumors are those that cause: Nadler-Wolfer-Elliot syndrome (hepatoma-hypoglycemia), Rosenfeld syndrome (pseudomyxoma-hypoglycemia), Andersen syndrome (renal tumor-hypoglycemia).

Increased alkaline phosphatase syndrome

Stauffer syndrome in hypernephroma.

ENDOCRINE PARANEOPLASTIC SYNDROMES

They consist of pathological, clinical or sometimes only biological manifestations, linked to an ectopic hormonal secretion by neoplastic cells.

Paraneoplastic endocrine syndromes are the best characterized and the criteria that define these conditions can be specified:

1. Increased hormone levels.
2. Reduction in hormone levels after tumor removal or treatment.
3. Persistence of hormonal elevation after removal of the normal organ that secretes the substance.
4. Hormonal arteriovenous gradient in the tumor vascular bed.

5. Demonstration that tumor cells in vitro synthesize and secrete this hormone.

Hormones may be elevated for several years before symptoms develop, and many tumors produce various hormones and more than one has biological activity.

Paraneoplastic endocrine syndromes develop in a minority of tumors, but they have the importance of being, on occasion, the first symptom of the disease and hormonal elevation can be used as a marker of relapse. The treatment of these syndromes is important to control the clinical manifestations of excess hormones and to prevent autocrine growth of the tumor.

The mechanisms of production of endocrine paraneoplastic syndromes are the following:

1. Production of active proteins such as hormones, growth factors, cytokines, oncofetal proteins (AFP), immunoglobulins or enzymes.
2. Immune mechanisms: autoimmunity, immune complexes and immune suppression.
3. Mechanisms of production of an "ectopic" receptor or blockage of normal hormones by inactive hormones produced by the tumor.
4. Mechanism of "forbidden contact" in which enzymes or other products are released that do not normally circulate, but that, due to the abnormal vascularization of the tumor or the alteration of the membranes, allow antigenic reactions or other toxic manifestations.

Paraneoplastic Cushing syndrome

It is the best individualized, the initial tumor has three preferred locations: the lung, the thymus and the pancreas.

Paraneoplastic hyperthyroid syndrome

It should be considered with caution; Only the case described by Hennen, of bronchial cancer with the presence of TSH activity, has been adequately proven. Tumors that contain trophoblastic cells (choriocarcinoma, hydatidiform mole, teratoma of the testicle) should also be evaluated as causes of this syndrome.

Syndrome of inappropriate antidiuretic hormone secretion (Schwartz-Bartter syndrome)

It is essentially seen in oat cell bronchopulmonary carcinoma (see this syndrome).

Precocious puberty and gynecomastia syndrome

They are rarer, but gonadotropic activity has been demonstrated in tumor tissue. They have been observed in liver neoplasms, breast cancer, malignant melanoma and feminizing adrenal tumor.

Ectopic prolactin secretion syndrome

It appears in rare cases of lung cancer and kidney cancer, and of three cases, only one presented galactorrhea, which was corrected with tumor resection.

Growth hormone syndrome

It has been detected in patients with lung, gastric and carcinoid cancers. It is debated whether hypertrophic osteoarthropathy is due to this mechanism.

Finally, we should point out that associations of endocrine syndromes due to the production of various hormones by tumor tissue have been described.

PARANEOPLASTIC SYNDROMES

CARDIOVASCULAR CONALTERATIONS

1. Varicocele syndrome with thrombophlebitis (hypernephroma).
2. Endocarditis syndrome (carcinoid tumor and others).
3. Severe thrombophlebitis syndrome with pulmonary embolism; acrocyanosis and acrogangrene, pseudoRaynaud (hemoblastosis)
4. Migratory thrombophlebitis syndrome, Trousseau syndrome.

PARANEOPLASTIC SYNDROMES

WITH DIGESTIVE MANIFESTATIONS

1. Hyperacidity syndrome, gastric and duodenal ulcers, Zollinger-Ellison syndrome (benign or malignant adenoma).
2. Pseudoileus colon syndrome, Ogilvie syndrome.
3. Depletion syndrome, with complex alterations in fat and protein digestion and hepatomegaly.
4. Stauffer syndrome (hypernephroma).

PARANEOPLASTIC SYNDROMES

HEMOLYMPHOPOIETIC

These syndromes involve hematological cells, immunoglobulins, and blood clotting.

Polyglobulia syndrome

It appears in 10-20% of patients with cerebellar hemangioma, in 1-5% of renal cancers and less frequently in hepatoma and adrenal tumors. In the majority of these tumors, an increase in erythropoietin is found, which, in general, does not produce clinical manifestations. Paraneoplastic erythrocytosis should be considered in the diagnosis of polyglobulia.

Eritoblastopenia syndrome

In half of the patients it is associated with thymoma. Autoimmune-based cases accompanied by antinuclear antibodies, paraproteinemia and autoimmune hemolytic anemia or, in rare cases, antierythropoietin antibodies, have been described.

Autoimmune hemolytic anemia syndrome

It appears more frequently in chronic lymphatic leukemia (10%) and to a lesser extent in ovarian cancer and lymphomas. Anemia affects a condition with the characteristics of extravascular regenerative hemolysis. Hemolysis is due to warm IgG type antibodies, direct Coombs positive, with cold IgM type antibodies being rarer.

Microangiopathic hemolytic anemia syndrome

It is a rare manifestation that is usually associated with mucin-producing adenocarcinomas, such as gastric cancer, or in patients with hemangiomas in which a traumatic injury is exerted on the red blood cells. It is characterized by a condition with anemia, schistocytosis and reticulocytosis, unconjugated hyperbilirubinemia, hemoglobinuria and siderinuria, and thrombocytopenia associated with disseminated intravascular coagulation.

Granulocytosis syndrome

An important group of reactions appear in tumors without bone marrow infiltration. The tumors in which it occurs most frequently are gastric cancer, bronchial cancer, pancreatic cancer, melanoma, Hodgkin's disease, some lymphomas and tumors of the nervous system. It is possible that the mechanism is related to the production of hematopoietic growth factors.

Eosinophilia syndrome

It is defined by an increase in eosinophils greater than 700/mL. It appears in 20% of patients with Hodgkin's disease, possibly due to the action of IL-5. It also appears in myeloproliferative syndromes and lung cancer. It is not associated with clinical manifestations related to eosinophilia.

Thrombocytosis syndrome

An increase in platelets greater than 400,000/mL appears in one third of cancers. In the diagnosis, the possibility of an inflammatory process, hemorrhagic episodes, iron deficiency and postsplenectomy must be considered. Thrombocytosis does not usually cause clinical complications and in a group of cases coincides with an increase in IL-6.

Disseminated intravascular coagulation (DIC) syndrome

The tumors most frequently associated with it are mucin-producing adenocarcinomas, such as gastric, pancreatic, lung, prostate, colon cancers, and acute promyelocytic leukemia. The clinical picture can present itself acutely and chronically and the most characteristic tests for diagnosis are the lengthening of the partial thromboplastin time, prothrombin and thrombin time, reduction in the number of platelets, presence of soluble fibrin monomers, etc.

Paraneoplastic syndrome associated with paraproteins

It can inhibit the aggregation of fibrin monomers, factor VIII and factor

PARANEOPLASTIC SYNDROMES THAT FUNDAMENTALLY AFFECT ONE ORGAN OR REGION

Paraneoplastic liver disease syndrome

It is characterized by alteration, sometimes reversible, of liver function tests without the existence of metastasis. There is elevation of alkaline phosphatase, hypocholesterolemia, lengthening of prothrombin time and hepatosplenomegaly, which disappears with removal of the tumor. It is more frequently associated with hypernephroma and is related to a possible amyloid deposit or hepatic hypervascularization. It is important to diagnose this condition and not confuse it with the existence of liver metastasis.

Anorexia and cachexia syndrome

A third of cancer patients have a negative nitrogen balance. The size of the tumor mass is related to the alteration of the clinical status and the intensity of the general manifestations. The consideration of paraneoplastic syndrome is evident when tumor resection corrects anorexia.

TNF-alpha and IL-1B produce a cachexia-like condition in animals. These cytokines inhibit lipoprotein lipase activity in peripheral tissues and lead to a metabolic disorder resulting in anorexia and cachexia. Small cell lung carcinoma, gastric cancer and pancreatic cancer are the tumors in which these manifestations occur with greater intensity.

Febrile syndrome

It coincides with certain tumors, with Hodgkin's disease, hypernephroma, hepatoma and osteogenic sarcoma. Fever occurs without another justifying cause, mainly infection, and disappears with removal or therapeutic control of the tumor. The cause of fever is the release of a pyrogen by the tumor (from the Kupffer cells and lymph nodes) that acts on the hypothalamus and causes temperature alteration.

Pulmonary hypertrophic osteoarthropathy syndrome

It is a condition characterized by clubbing of fingers, periostitis of the long bones and, sometimes, polyarthritis, reminiscent of rheumatoid arthritis. This condition usually precedes the diagnosis of the tumor by several months. It appears almost selectively in lung adenocarcinoma, with a frequency of 12% and is very rare in small cell lung cancer.

Paraneoplastic nephropathy syndrome

Only glomerular lesions and obstruction by tumor products should be considered as such. The glomerular paraneoplastic condition is based on a nephrotic syndrome with massive proteinuria. Hodgkin's disease is the most common tumor associated with this condition and to a lesser extent non-Hodgkin's lymphomas of intermediate and high degrees of malignancy are accompanied.

The renal lesions that appear in carcinomas are similar to membranous glomerulonephritis induced by immune complexes in animals. This suggests a deposition of circulating antigen-antibody complexes in the renal glomerulus. Lipoid nephrosis in patients with Hodgkin's disease derives from a deficit in the function of T lymphocytes and in non-Hodgkin's lymphomas, immunoglobulin deposits are identified that suggest a deposition of immune complexes.

AARSKOG SYNDROME

Synonymy

Facial-digital-genital syndrome.

Syndromography

Clinic

This syndrome is manifested by low height with a postnatal onset (between 2-4 years) that reaches less than the third percentile, rarely reaching a height of 160 cm in adults. It is characterized by dysmorphism of the face, hands and genitals.

Physical examination. Facies: presents a wide forehead with prominent metopic suture, ocular hypertelorism, anti-mongoloid deviation of the palpebral fissures and ptosis of one or both upper eyelids, short and wide nose with upward nostrils, the middle region of the face is somewhat flattened, the pliltrum is elongated (nasolabial distance), the upper lip is thick with a linear groove below the lower lip. The earlobes are usually thick and the upper helix poorly formed. Hands: are short and wide (brachydactyly) with alenodactyly of the fifth finger, hyperextensibility of the proximal interphalangeal joints that give an unusual position to the extended fingers, simian groove and increased interdigital membrane.

Genitals: The scrotal fold extends around the base of the penis, giving the impression of a saddlebag scrotum.

Other clinical manifestations are: moderate pectus excavatum, prominent umbilicus, inguinal hernias, slight mental retardation, cryptorchidism, hypodontia, delayed dental eruption.

Paraclinical examinations

Radiology. Delayed bone age, hypoplasia of the first cervical vertebra with subluxation or fusion of the first cervical vertebrae. Hypoplasia of the distal and middle phalanges.

Syndromogenesis and etiology

It is a syndrome of monogenic genetic etiology of recessive inheritance linked to the chromosome with expression in heterozygous women, especially on the

Evidence of autosomal dominant inheritance has been reported, so genetic heterogeneity could be raised. The basic defect is not known.

Literature

Aarskog, D.: A familial syndrome of short stature associated with facial dysplasia and genital anomalies". J. Pediatr., 77:856-861, 1970.

Berman, P.; C. Desjardins and F.C. Fraser: "The inheritance of the Aarskog syndrome." J. Pediatr., 86:885-891, 1975.

Grier, C, et al.: "Autosomal dominant inheritance of the Aarskog syndrome." Am. J. Med. Genet., 15:39-46, 1983.

AASE-SMITH SYNDROME

Synonymy

Arthrocerebro-palatine dysmorphogenesis. S. of triphalangeal thumbs-anemia.

Syndromography

Clinic

Chronic non-progressive condition that begins in intrauterine life (can be fatal), is present at birth in its complete or incomplete forms. It affects both sexes. Mental retardation (not constant).

Physical examination. This syndrome is poorly defined but is characterized by a size around the 3rd. percentile, triphalangeal thumbs, moderate radial hypoplasia, narrow shoulders, late closure of the fontanels. Severe non-progressive joint contractures, hip dislocation, absence of flexion folds and fingerprints (dermatoglyphs), cleft palate, malformed ears, hydrocephalus, oculomotor paralysis. Congenital heart disease, especially septal defect. Hepatos- variable plenomegaly.

Paraclinical examinations

Blood. Absence of biochemical alterations. Hypoplastic anemia and variable leukopenia. The anemia responds to treatment with prednisone and improves with age.

Chromosome analysis. Normal.

Radiology. The described alterations are confirmed.

Syndromogenesis and etiology

The occurrence in siblings of both sexes suggests a genetic etiology of autosomal recessive inheritance.

Literature

Aase, J.M. and D.S. Smith: "Congenital anemia and triphalangeal thumbs. A new syndrome." J. Pediatr., 74: 471-474, 1969.

Muis, N. et al.: The Aase syndrome: case report and review of the literature'". Europ. J. Pediat., 145:153-157, 1986.

ACHARD SYNDROME

Syndromography

Clínica

This syndrome is characterized by presenting an external habit similar to Marfan syndrome, without alteration of body proportions and the height is not excessively tall. He presents arachnodactyly, a wide skull with brachycephaly, microretrognathia and joint hypermobility limited to the hands and feet.

Paraclinical examinations

Those necessary to rule out Marfan syndrome (see s. of Marfan).

Syndromogenesis and etiology

It is not well defined if it is different from Marfan syndrome, since they only seem to differ in craniofacial dysmorphism.

Literature

Achard, C.: "Arachnodactyly." Bull. Mem. Soc. Med. Hosp., Paris. 19:834-840, 1902.
Parish, J.G.: "Heritable disorders of connective tissues with arachnodactyly." Proc. Roy. Soc. Med., 53:515-518, 1960.

ISOVALERIC ACIDEMIA SYNDROME

Syndromography

Clínica

This syndrome occurs in both sexes. The newborn is normal at the time of birth but after a few days he suffers from attacks of vomiting, acidosis, coma, a strong body odor (sweaty feet smell), which increases with the recurrence of symptoms, medium psychomotor retardation and aversion to foods rich in proteins. Neonatal death may occur. The treatment is based on a diet low in leucine.

Paraclinical examinations

Blood. Accumulation of isovaleric acid. Ketonemia during crises.

Urine. Excretion of isovaleric acid (typical odor). Leucine tolerance test.

Syndromogenesis and etiology

Leucine metabolism error with accumulation of isovaleric acid in serum. Isovaleryl-CoA- -dehydrogenase defect.

It is apparently transmitted by autosomal inheritance. recessive.

Literature

Cruz, F.: Treaty of Pediatrics, t.I, v.I, p.351, 1983.
Budd, M.A.: K. Tanaka; L.B. Holmes et al.: Isovaleric acidemia". New Eng. J. Med., 277:321-327, 1967.

FAMILY INFANTIL LACTIC ACIDOSIS SYNDROME

Synonymy

S. of congenital lactic acidosis. S. of congenital chronic lactic acidosis.

Syndromography

Clínica

It begins in the first year (sometimes in newborns) and is characterized by: tachypnea, muscle hypotonia, attacks of intense lactic acidosis (very high lactoacidemia). In the following months, brain deterioration, ataxia, seizures or tremors, and psychomotor retardation appear due to Leigh's necrotizing encephalopathy. Abnormal eye movements. Partial alopecia. Death usually occurs before the age of 2 years in two-thirds of cases.

Paraclinical examinations

Blood. Hyperalaninemia and hypophosphatemia. Very high lactic acid.

Urine. Aminoaciduria.

Syndromogenesis and etiology

The mechanism appears to be a defect in pyruvate decarboxylation. It is transmitted by autosomal recessive inheritance.

The etiology is unknown.

Literature

Hartmann, A.F.: H.J. Moltmann: J. Purkerson et al. "Lactate metabolism. Studies of a child with a serious congenital deviation" J. Pediat., 61:165, 1962.
Erickson, R.J.: "Familial infantile lactic acidosis". J. Pediat.. 66:1004, 1965.

PROXIMAL RENAL TUBULAR A CIDOSIS (ART) SYNDROME

Synonymy

S. de Rodríguez-Soriano-Edelman. S. of ART type "bicarbonate waste." S. of ART "transitory" form.

Syndromography

Clinic

It is observed in boys and generally begins before the age of one and a half due to anorexia and vomiting, especially at the beginning.
Slow growth with percentiles below the weight and height percentile. Bone immaturity. He did not present nephrocalcinosis, rickets, osteomalacia, or pathological fractures.

Paraclinical examinations

Blood. Hyperchloremic acidosis. Low pH and bicarbonate, chloremia greater than 110 mEgL.

Urine. pH greater than 6. High bicarbonaturia in the presence of mild acidosis. When exploring tubular function through bicarbonate perfusion, the existence of a threshold and a reduced maximum transport capacity are verified. Urine may be more acidic if plasma bicarbonate levels are low enough to allow complete reabsorption. Ammonium chloride overload.

Radiology. Bone immaturity.

Kidney biopsy. No specific injuries have been described.

Syndromogenesis and etiology

This syndrome is caused by depression of the renal threshold for bicarbonate, which causes metabolic acidosis. It is suggested that this acidosis may be due to a transient abnormality of bicarbonate reabsorption in the proximal tubule, since it responds to the intense and prolonged administration of mixed sodium and potassium citrate or bicarbonate; while the distal mechanisms of hydrogen ion excretion are normal. The loss of bicarbonate leads to a decrease in the plasma alkaline reserve, which would cause severe acidosis if it were not for the functional integrity of the most important distal mechanisms of acid-base balance regulation.

It is transmitted by sex-linked recessive inheritance, it appears only in males.

The etiology is genetic.

Literature

Lightwood, R.: N.E. MacLagan and J.G. Williams: Persistent acidosis in an infant cause not yet ascertained." Proc. Roy. Soc.Med.. 29:1431, 1936.

Rodríguez-Soriano, J.; H. Boichis and H. Stark: "Proximal renal tubular acidosis: A defect in bicarbonate reabsorption with normal inay acidification. Pediat. Res.. 1:81, 1967.

Nash, A.D. Roasted; I. Greifer and colbs: *Renal tubular acidosis in infants and children". J. Pediat., 80:738, 1972.

METHYLMALONIC ACIDURITY SYNDROME

Synonymy

See hereditary syndromes of amino acid metabolism.

Syndromography

Clinic

Both sexes, from birth. Persistent vomiting, lethargy. If they survive, developmental disorders, mental retardation.

Paraclinical examinations

Blood. During attacks: neutropenia, thrombocytopenia, ketoacidosis, hyperammonemia, hypoglycemia, intermittent hyperglycinemia.

Urine. Hypoxaluria, intermittent hyperglycinuria and massive excretion of methylmalonic acid.

Syndromogenesis and etiology

It is transmitted by autosomal recessive inheritance. There is difficulty in metabolizing C-propionate to succinate, which causes an accumulation of methylmalonate.

The etiology is unknown.

Literature

Barness, L.A. and G. Morrow II: "Methylmalonic aciduria. A newly discovered inborn error." Ann. Int. Med., 69:633-635. 1968.

Oberholzer, V.G.; B. Levin; E.A. Burgess et al.: "Methylmalonic aciduria. Ann inborn error of metabolism leading to chronic metabolic acidosis'." Arch. Dis. Child., 42: 492-504, 1967.

HEREDITARY OROTIC ACIDURIA SYNDROME

Synonymy

S. of orotic aciduria.

Syndromography

Clinic

Predominates in men. There are no abnormalities at birth. During the first year, there is developmental difficulty, apathy, a certain degree of mental retardation, lassitude, and repeated infections.

Physical examination. Paleness, muscle hypotonia, no neurological alterations.

Paraclinical examinations

Blood. Hypochromic anemia with marked anisopoikilocytosis.

Urine. Excessive excretion of orotic acid.

Bone marrow. Atypical megaloblastic changes.

Syndromogenesis and etiology

Enzymatic defect with disorder in the conversion of orotic acid to uridine-5-phosphate, and overproduction of orotic acid.

It is transmitted by autosomal recessive inheritance. The enzymatic defect is deficiency of orotidine-5-primaphosphate decarboxylase.

Literature

Huguley, C.M., Jr.; J.A. Bain; S.L. Rivers et al.: "Refractory megaloblastic anemia associated with excretion of orotic acid." Blood, 14:615-634, 1959.

Smith, L.H.; C.M. Huguley, Jr. And J.A. Bain: Hereditary orotic aciduria. In Stanbury, J.B., J.B. Wyngaarden, and D.S. Frederickson: The metabolic basis of inherited disease. ed. 2,. McGraw-Hill, New York, 1966.

ACHONDROGENESIS SYNDROME TYPE I

Synonymy

S. Parenti-Fraccaro type (type IA and IB). Houston-Harris. Type IA. Fraccaro type IB.

Syndromography

Clinic

This syndrome is currently classified into two types: IA and IB.

Clinically, both patients present extreme short stature from prenatal onset. They are born with a size between 22 and 30 cm. The skull is large for gestational age, the nasal bridge depressed, micrognathia. The limbs are extremely short like fins.

In the prenatal history, there are frequent stories of polyhydramnios, dropsy, and fetal death. The majority are stillborn newborns or die shortly after birth.

Paraclinical examinations

Radiology. It is specific for each type, but in both there are common elements: the skull, vertebral bodies, fibula, talus and calcanei are poorly ossified, the iliacus are toothed, the long bones stellate and the ribs extremely short.

In type IA there are multiple rib fractures and the proximal metaphysis of the femur has spikes.

In type IB there are no rib fractures and the distal metaphysis of the femur is irregular and the femur is trapezoid.

Syndromogenesis and etiology

The basic defect is not known, however it seems to be related to defects in the connective tissue of cartilage and bone. In type IA there is hypervascularity of the cartilaginous matrix and increased cell density, the chondrocytes contain round cytoplasmic inclusion bodies and are surrounded by a large lacuna. In type IB there is a marked deficiency of collagen fibers, the chondrocytes are large with a round central nucleus and surrounded by a dense ring of collagen.

The etiology is monogenic genetics of autosomal recessive inheritance.

Literature

Whitley, C.B. and R.J. Garlin: “Achondrogenesis: New nosology with evidence of genetic heterogeneity.” Radiology. 148:693-698, 1983.

ACHONDROGENESIS SYNDROME TYPE II

Synonymy

S. de Langer-Saldino.

Syndromography

Clínic

It is classified as bone dysplasia of prenatal onset, with extremely small size (27-36 cm). It is characterized by a large skull with large fontanelles, a depressed nasal bridge, anteverted nostrils, and micrognathia. Very short limbs and occasionally cleft soft palate. Most are stillborn or die a few hours after birth.

Prenatal history of polyhydramnio.

Paraclinical examinations

Radiology. Normal skull ossification. Short ribs without fractures. Long, short and wide bones, disproportionately long fibula (fibula), metaphysical irregularities in the distal portion of the ulna (radius). Ossification defects of the lumbar, cervical, sacral, pubic, calcaneal and talus bones.

Syndromogenesis and etiology

Monogenic genetic etiology of autosomal recessive inheritance. The basic defect is related to a mutation in exon 46 of the type II collagen gene, changing the glycine codon to the serine codon (GAC to AGC) at position 943;
There is also abnormality of cartilage protoglycans, but it is unclear whether this is a primary or secondary finding. Histology reveals hypervascularity and hypercellularity of the cartilage with multiple small dilated cisterns of rough endoplasmic

Literature

Chen, H.; C.T. Lin and S.S. Yang: “Achondrogenesis a review with special consideration of achondrogenesis type II (Langer-Saldino)”. Am. J. Med. Gent., 10:379-394, 1981.

Vissing; H. et al.: "Glycine to serine substitution in the triple helical domaine of pro 1 (I) collagen results in a lethal perinatal form of short-limked dwarfism." J. Biol. Chem., 264: 18265-18268, 1989.

ACROCEPHALOSINDACTYLY TYPE I SYNDROME (APERT)

Synonymy

S. of Apert. S. of syndactyic oxycephaly.

Syndromography

Clinic

It is characterized by irregular craniosynostosis, hypoplasia of the middle third of the face, and severe syndactyly.

Craniofacial alterations: shortening of the anteroposterior diameter of the skull and increase in its height, high forehead and flat occiput. Craniosynostosis of the coronal suture although it is irregular. Large late-closing fontanels. Flattened facies, small orbits, with horizontal supraorbital groove, hypertelorism, antimongoloid deviation of the palpebral fissures, strabismus, small nose, maxillary hypoplasia. Narrow palate, with or without cleft palate or bifid uvula.

Hands: bone and skin syndactyly that varies from total or partial fusion, commonly with complete fusion of the second, third and fourth fingers. The distal phalanges of the thumbs are often wide and in a valgus position, the fingers may be

Short.

Feet: cutaneous syndactyly with or without bony syndactyly.

There may be limitation of joint mobility.

tions.

Severe to moderate acne is described in adolescents.

They may present mental retardation but also normal intelligence.

Paraclinical examinations

Radiology. The described alterations are confirmed, there may be short humeri, radio-ulnar synostosis. Increased digital impressions on the skull.

Cerebrospinal fluid. Elevated endo-pressure

cranial.

Syndromogenesis and etiology

The etiology is monogenic genetics with autosomal dominant inheritance. Almost all cases represent new mutations, with advanced paternal age being an important factor.

The mutant gene may adversely affect the organization of bone development and other tissues, especially in the distal limbs and skull.

Literature

Apert, E.: “De laccrocephaalosyndactyly”. Bull. Soc. Med. Paris, 3s, 23:1310-1330, 1906.

Buckanan, R.C.: "Acrocephalosyndactyly or Apert's syndrome." Br. J. Plast. Surg., 21:406-418, 1968.

Cohen, M.M.: “An etiologic and nosologic overview of cranio-synostosis syndrome.” Birth Defects., 11 (2): 137-189, 1973.

Skoog, T.: "Syndactyly, a clinical report on repair." Chir Act. Scand.. 130:537-549, 1965.

ACROCEPHALOPOLISINDACTYLY TYPE II SYNDROME (CARPENTER)

Synonymy

Carpenter's S. S. of cephalodactyly. by Apert-Crouzón.

Syndromography

Clinic

It is characterized by acrocephaly, polysyndactyly and displacement of the internal canthi.

Craniofacial alterations: brachycephaly with variable synostosis of the coronal, sagittal and lambdoid sutures. Narrow supraorbital rings. Lateral displacement of the inner canthus of both palpebral fissures with or without epicanthus.

Hands: brachydactyly of the hands with clinodactyly, partial syndactyly (third and fourth fingers) and camptopdactyly.

Feet: preaxial polydactyly with partial syndactyly.

Coxa valga: with baro foot.

Other defects: accessory spleen, abdominal hernias, short stature, kidney anomalies and sensorineural deafness. They may present mental retardation.

Presence of congenital heart disease. Hypoge-

nitalism.

Upper jaw hypoplasia

Paraclinical examinations

Radiology. The described anomalies can be seen, flattening of the acetabulum, coxa valga, lateral displacement of the patella.

Abdominal ultrasound, hearing tests, computed axial tomography. They can be useful for the diagnosis of kidney and spleen anomalies, as well as for the study of hearing defects.

Syndromogenesis and etiology

It is assumed that the defect has a genetic etiology due to a monogenic autosomal recessive mutation, but there is no known evidence that explains the defects of the syndrome.

Literature

Carpenter, G.: Two sisters showing malformation of the skull and other congenital abnormalities." Rep. Soc. Study Dis. Child, London, 1:110-118, 1901.

Eaton, A.P. et al.: "Carpenter's syndrome acrocephalopolysyn dactyly type II." Birth Defects, 10(9):249-260, 1974.

Temtamy, S.A.: "Carpenter's syndrome acrocephalopolysyndactyly: An autosomal recessive syndrome." J. Pediatr., 69:111- -120, 1966.

ACROCEPHALOSINDACTYLY TYPE II SYNDROME (CHOTZEN)

Synonymy

S. de Saethre-Chotzen. S. de Chotzen.

Syndromography

Clinic

The most relevant clinical characteristics of this syndrome are: brachycephaly with maxillary hypoplasia, prominence of the crux of the helix and cutaneous syndactyly.

Craniofacial alterations can be summarized as brachycephaly with a high forehead due to synostosis of the coronal suture. There is hypoplasia of the maxilla with a narrow palate, facial asymmetry with deviation of the nasal septum, narrow orbits, hypertelorism, eyelid ptosis, dysplastic ears with prominence of the root of the helix. There may be some degree of loss of hearing acuity. The fontanels are large and close late.

Patients may have normal intelligence, slight to moderate degree of mental deficiency, short stature, increased intracranial pressure.

In the hands they present partial cutaneous syndactyly frequently between the second and third fingers that can extend to the fourth finger, brachydactyly with small distal phalanges and clinodactyly of the fifth finger. The feet are characterized by a wide first joint, hallux valgus.

Paraclinical examinations

Radiology. The bone alterations described are confirmed; the clavicles may also be shortened or hypoplastic; vertebral anomalies.

Syndromogenesis and etiology

The etiology is genetic due to a monogenic mutation that is transmitted with autosomal dominant inheritance with a wide range of variability in its phenotypic expression.

The relationship between craniosynostosis and mental development remains uncertain.

Literature

Bartsocas, C.S.: A.L. Weber and J.D. Crawford: Acroce- phalosyndactyly type III: Chotzen's syndrome'". J. Pediatr., 77:267-269, 1970.

Friedman, J.M. et al.: "Saethre-Chotzen's syndrome: a broad and variable pattern of skeletal malformations." J. Pediatr., 91:929, 1977.

PFEIFFER TYPE ACROCEPHALOSINDACTYLY SYNDROME

Synonymy

S. de Pfeiffer.

Syndromography

Clínica

Brachycephaly with craniosynostosis of the coronal, moderate of the sagittal with high forehead, ocular hypertelorism, antimongoloid deviation of the palpebral fissures, small nose with depression of the nasal bridge, narrow maxilla.

Hands and feet: distal phalanges, thumb and big toe wide, middle phalanges of the fingers somewhat small, partial syndactyly of the second and third fingers of the hands and of the second, third and fourth toes.

Other anomalies may occur with choanal atresia, cloverleaf skull, mental retardation, hydrocephalus, seizures.

Intelligence is usually normal.

Paraclinical examinations

Radiology. Craniosynostosis is confirmed: there may be radio-ulnar synostosis of the elbow.

Syndromogenesis and etiology

The etiology is genetic, the mutant gene is transmitted as an autosomal dominant inheritance.

The basic defect that explains the clinical manifestations is unknown.

Literature

Naveh, Y. and A. Friedman: "Pfeiffer's syndrome: a report of the family and review of the literature." J. Med. Genet., 13:277- -280, 1976.

ACHRODYSOSTOSIS SYNDROME

Synonymy

S. of peripheral dysostosis.

Syndromography

There are few cases described in the literature.

Clinic

Moderate low stature from prenatal onset. Between 35-85% of reported cases present mental retardation.

Craniofacial alterations: brachycephaly, depressed nasal bridge with little development of the nasal bones, small nose

ña with anteversion of the nostrils. Tendency to keep the mouth open. Maxillary hypoplasia with prognathism.

There may be dental malocclusion, hydrocephalus, recurrent ear infections, optic atrophy, hypogenitalism, hypogonadism.

The limbs are short, especially in the distal regions, the hands appear short and wide with wrinkling of the skin on the back.

Paraclinical examinations

Radiology. Brachycephalic skull with occasional hyperostosis, progressive distal deformity of the humerus, radius and ulna. Epiphyses of the phalanges in the form of cones that fuse prematurely into the hands and feet.

Syndromogenesis and etiology

Cases occur sporadically.

The etiology is unknown.

Literature

Robinow, M. et al.: "Acrodysostosis. A syndrome of peripheral dysostosis, nasal hypoplasia, and mental retardation." Am.J. Dis. Child., 121: 195-203, 1971.

ADDISON-SCHILDER SYNDROME

Synonymy

Addison's-cerebrosclerosis. Addison's syndrome-diffuse cerebral sclerosis. S. of adrenoleukodystrophy.

Syndromography

Clinic

Cerebral sclerosis associated with Addison's disease is a very rare form of recessive inheritance linked to sex and appears only in men. In these patients, a picture completely identical to Schilder's cerebral sclerosis develops, as well as a typical clinical form of Addison's disease. The relationship between endocrine disease and neurological disorders justifies the name adrenoleukodystrophy.

The terms melanodemic adrenoleukodystrophy or sudanophilic leukodystrophy have also been used.

It begins between 3-10 years, the skin is hyperpigmented, brown, as well as the oral mucosa and scar areas. These alterations precede by years the neurological manifestations that begin between 5-15 years of age, and are characterized by: behavioral and gait alterations, weakness of one or several limbs, which progresses to spastic quadriplegia with hyperreflexia and Babinski, seizures, decortication posture, stupor, deterioration of speech, dysarthria and intelligence, and finally dementia. Dysphagia and visual defect with vision loss.

After the age of 4, symptoms and signs of adrenal insufficiency appear and skin pigmentation becomes accentuated.

Paraclinical examinations

Blood. Hypoglycemia.

Urine. Decrease in 17-ketosteroids without response to ACTH.

Cerebrospinal fluid. Hyperalbuminorrhachia.

Syndromogenesis and etiology

In patients with this syndrome there is an inability to oxidize very long chain fatty acids. They have been demonstrated levels of very long chain saturated fatty acids, especially hexicosanoic acid (C26) in plasma, adrenal cortex, cerebral white matter and in cultures of fibroblasts from the skin and amniotic fluid.

This syndrome demonstrates hypoplasia that leads to atrophy of the adrenal cortex. Marked demineralization of the white matter of the cerebral hemispheres, with infiltration of lymphocytes and macrophages loaded with fat.

It is transmitted by sex-linked autosomal recessive inheritance.

It is possible to identify heterozygous females in families with affected children and prenatal diagnosis has been successfully carried out.

The etiology is genetic.

Literature

Fanconi, A.; A. Prader: W. Isler et al.: "Morbus Addison mit Himsklerose im Kindesaliter: Ein hereditaires Syndrome mit- X-chromosonmaler Vererbung?" Helv. Paediat. Acta, 18:480, 1964.

Siemerling, E. und H.G. Creutzfels: "Bronzkrankhent und skelorosierende Enzephalomyelitis (Diffuse Sklerose)". Arch. Psychiat. Nervenkr., 68:217, 1923.

Turlington, R.W. and R.S. Stempfel: "Adrenocortical atrophy and diffuse cerebral sclerosis (Addison-Schilder disease)." J. Pediat., 69:406, 1966.

CEREBELLAR VERMIS AGENESIA SYNDROME

Synonymy

S. de Joubert. S. de Joubert-Boltshauser.

Syndromography

Clinic

Symptoms may begin in childhood in the form of tremor and hypotonia. Other common symptoms are: hyperpnea, abnormal eye movements and psychomotor delay. The oldest patient lived to be 8 years old. Sometimes the signs and symptoms are related to obstruction of the ventricular system:

headache and intracranial hypertension. Other malformations have been reported: chorioretinal coloboma, Dandy-Walker type malformation, hypoplasia of the corpus callosum, occipital meningoencephalocele, bilateral optic nerve

Paraclinical examinations

Computed tomography. It is more useful and less risky than the pneumoencephalogram.

Syndromogenesis and etiology

It is a genetic syndrome with autosomal recessive inheritance, due to consanguinity observed in parents of affected children.

A syndrome of aplasia of the cerebellar vermis has been described that presents non-progressive ataxia from birth and sometimes vertical nystagmus. The diagnosis is made by magnetic resonance imaging. It is a genetic syndrome and autosomal dominant, or X-linked dominant, inheritance is suggested.

Literature

Dow, R.S. and G Moruzzi: The Physiology and Pathology of the Cerebellum. Minneapolis, University of Minnesota Press, 1958.

Fenichel, G.M. and J.A. Phillips: "Familial aplasia of the cerebellar vermis possible X-linked dominant inheritance". Arch. Neurol. 46:582-583, 1989.
Joubert, M. et al.: "Familial agenesis of the cerebellar vermis. A syndrome of episodic hyperpnea, abnormal eye movements, ataxia and retardation. " Neurology, 19:813-825, 1969.
Van Dorp, et al.: "Joubert's syndrome: a clinical and pathological description of an affected male and a female fetus from the same sibship." Am. J. Med. Genet., 40:100-104, 1991.

AGLOSIA-ADACTYLY SYNDROME

Synonymy

S. of hypoglossia-hypodactyly.

Syndromography

Clinic

Facies: bird face with hypoplasia of jaw and lower lip.

Mouth: total or only partial lack of the tongue, especially the anterior portion. Microglossia has been described.

Ankyloglossic bands that fix the lingual rest to the posterior surface of the lower lip, passing over the alveolar edge, favored by the hypodontia of the lower incisors. However, speech articulation is not greatly affected unless the absence of the tongue is significant. High palate sometimes fissured.

Limbs: defects range from peromelia of several segments to just the absence of some terminal fingers:
partial anonychia of the fingers. Syndactyly.

Other occasional anomalies: situs inversus viscerum, dextrocardia, hyperplasia of submandibular glands. Fusion of large vulvar lips not constant. When there are brain defects, especially in the nuclei of the cranial nerves, it is accompanied by a Moebius sequence.

Intelligence and height are normal. The chromosome study is normal. It resembles other syndromes, but in Hanart syndrome there is no aglossia.

Syndromogenesis and etiology

It is due to an intrauterine disruptive defect caused by vascular defects that affect the distal regions: limbs, tongue and occasionally parts of the brain.

The etiology is unknown, generally the syndrome occurs sporadically.

Literature

Kelln, E.E.; DC Bennett and W.G. Killinger: “Aglossia-Adactyly syndrome.” Am. J. Dis. Child., 116:549, 1968.
Rossenthal, R.: "Aglossia congenital. A report of a case of the condition with other congenital malformations." Am. J. Dis. Child.. 44:383. 1932.

ALAGILLE SYNDROME

Synonymy

S. of ductal hypoplasia. S. of arteriohepatic dysplasia.

Syndromography

Clinic

It occurs in both sexes with itching from the sixth month of life. Cholestasis of early onset, with moderate jaundice and hypocolic stools.

Physical examination. Constant hepatomegaly, of normal or firm consistency; Frequent splenomegaly and xanthomas that can be located on the extension surface of the fingers and other areas of the body.

Characteristic facies from birth, more accentuated from the first year of life: prominent forehead, sunken eyes with slight hypertelorism, saddling of the nasal root and small, pointed chin.

Cardiovascular anomalies: the most common is hypoplasia or stenosis of the pulmonary artery, which is well tolerated.

Vertebral anomalies: images of a non-welded anterior arch, with the appearance of spina bifida in the dorsal vertebrae that give the appearance of winged vertebrae, without scoliosis. Growth delay, moderate mental retardation, hypogonadism. High pitch voice. Post-embryotoxon determined by ophthalmological examination with a slit lamp.

Paraclinical examinations

Blood. Moderate hyperbilirubinemia, hypercholesterolemia with hyperlipidemia. Alkaline phosphatase increased 3-5 times its value. Negative viral antigens.

Liver biopsy. Significant reduction in the number of bile canaliculi, with an evident decrease in portal spaces. Moderate portal fibrosis.

Syndromogenesis and etiology

Genetic syndrome possibly of autosomal dominant transmission (some recessive cases).

Literature

Alagille, D.; M. Odrievre; M. Gautier and J.P. Dommergues: "Hepatic ductular hypoplasia associated with characteristic facies, vertebral malformations, retarded physical, mental and sexual development and cardiac murmur." J. Pediat., 86:63, 1975.
Greenwood, R.D.; A. Rosenthal; B.C. Crocker and colbs.: "Syndrome of intrahepatic biliary dysgenesis and cardiovascular malformations." Pediatrics, 58:243. 1976.
Henriksen, N.T.; F. Lansgmark; S.J. Sorland et al.: "Hereditary choletasis combined with peripheral pulmonary stenosis and anomalies." Pediat Act. Scand., 66:7, 1977.

ALBERS-SCHÖNBERG SYNDROME

Synonymy

S. of osteopetrosis. Autosomal recessive lethal osteopetrosis S. Marble disease. S. of familial osteopetrosis. S. of marble bone.

Syndromography

Clinic

Increased bone density is the fundamental disorder resulting in skeletal fragility. It starts very early from the fetal stage. Fragility predisposes to fractures that can begin at birth. In the first year of life, the head becomes voluminous, square in shape with a prominent forehead. The decrease in the lumen of the exit foramina of the cranial nerves determines alterations of these, especially of the optic nerve. Vision progressively decreases, there is eyelid ptosis, strabismus, optic atrophy, cataracts, retinal degeneration.

There is also progressive deafness and facial paralysis. Teeth develop poorly and tend to fall out. Hepatosplenomegaly and enlargement of the lymph nodes. The patients have short stature.

Paraclinical examinations

Radiology. Generalized bone sclerosis of the entire skeleton. Transverse bands in the metaphyseal regions and longitudinal striae in the diaphyses. The vertebrae show prominent anterior vascular notches. In advanced stages, the proximal part of the humerus and distal part of the femur adopt the configuration of a bottle.

Biochemical determinations. Hypophosphatemia and hypocalcemia.

Hematological studies. Hypochromic anemia or pancytopenia.

Syndromogenesis and etiology

The etiology is genetic of autosomal recessive inheritance with consanguinity in many families.

The basic defect is unknown although several theories have been proposed: hypersecretion of thyrocalcitonin, defective bone formation poor in fibrils. The condition is rare but its frequency is higher in Costa Rica. There is a form with autosomal dominant inheritance that has a more benign character.

Literature

Albers-Schönberg, H.: "Roentgenbilder einer seltenen Knochenerkranskung." Munchen. Med. Wschr., 51:365, 1904.

Bollerslev, J.: “Autosomal dominant osteoporosis: bone metabolism and epidemiological, clinical and hormonal aspects.” Endocr. Rev., 10:45-67, 1989.

Loria-Cortés, R.: E. Ouesada-Calvo andC. Cordero-Chaverri: "Osteopetrosis in children: a report of 26 cases." J. Pediatr., 91: 43-47, 1977.

Marks, S.C. Jr.: "Osteoporosis-multiple pathways for the interception of osteoclast function." Appl. Pathol., 5:172-183, 1987.

CONGENITAL ALKALOSIS SYNDROME OF RENAL ORIGIN

Syndromography

Clinic

Poor weight gain, vomiting, constipation, weakness.

Paraclinical examinations

Blood. Hypochloremic alkalosis with severe hypokalemia vera.

Urine. Alkaline. Increased excretion of chlorine and potassium. Aldosterone may be increased.

Stool. Normal excretion of chlorine and potassium.

Syndromogenesis and etiology

It is considered a congenital condition.

Literature

Fashena, G.J. and R.J. Martin: “Congenital alkalosis of renal origin." Amer. J. Dis. Child.. 79:1127-8. 1950.

GASTROINTESTINAL ALLERGY SYNDROME TO MILK

Synonymy

S. allergy to cow's milk.

Syndromography

Clinic

This syndrome begins in the newborn from 2 days to 4-5 months if they are fed cow's milk. It is characterized by vomiting, diarrhea with mucus and positive occult blood in the stool. It varies in intensity, from minimal to fulminant with digestive bleeding and collapse. Cramping occurs for months preceding diarrhea.

Physical examination. Delayed growth. It is accompanied by paleness, dehydration. Hypotension and tachycardia and sometimes shock after ingesting cow's milk. With appropriate treatment, patients remain asymptomatic. And frequently the allergy decreases or disappears after 2 years.

Paraclinical examinations

Sigmoidoscopy. It shows the mucosa of the rectum and colon that is red and has superficial ulcerations.

Biopsy. Shows lymphocytic infiltrate, plasma cells and granulocytes in the ulcerated areas of the mucosa and abscess formation at that level.

Cow's milk ingestion test. Must be done carefully

Lactose tolerance. Normal.

Stool. They contain antibodies that precipitate cow's milk. Other sugar intolerances must be ruled out to establish the differential diagnosis.

Syndromogenesis and etiology

Allergy to cow's milk. History of allergies or digestive conditions in the family.

Literature

Gryboski, J.D.: "Gastrointestinal milk allergy in infants." Pediatrics, 40:354-362, 1967.

Schols, O.M. and T.W. Whorthen: "The permeability of the gastroenteric tract of infants to indigest protein", Amer. J. Dis. Child, 11:342, 1916.

ALLAN SYNDROME

Synonymy

S. arginino-succinic-aciduria.

Syndromography

Clinic

There are two clinical forms:

1. Late form, the most common (78%), which begins in the second year of life. It is characterized by psychomotor retardation and seizures (40%), irritability and transient ataxia attacks of choreiform type and nystagmus that last a few days.

Delayed development, trichorrhexis nodosa in hair that is scarce and grows little, the hair is dry, brown, dull and very fragile; Under the microscope it presents knots. Hepatomegaly.

2. Early form (27%), malignant, with vomiting from the first days, food refusal, hepatomegaly. Apathy, drowsiness, coma and seizures, with death in the first weeks of life.

Paraclinical examinations

Urine. Large amount of argininosuccinic acid.

Blood and cerebrospinal fluid. Postprandial hyperammonemia.

Syndromogenesis and etiology

The astrocytes of the nervous system are transformed in type II Alzheimer's; toxic lesions in the liver and kidneys due to ammonemia. This is due to a deficiency of argininosuccinase that allows the accumulation of argininosuccinic acid and ammonia.

The low proportion of arginine damages the hair.

It is transmitted by autosomal recessive inheritance.

The etiology is genetic.

Literature

Allan, J.D.; D.C. Cusworth; EC. Dent et al.: "A diseases probably hereditary characterized by severe mental deficiency and a constant gross abnormality of amino acid metabolism." Lancet, 1:182, 1958.

Ferriaux, J.P.; C. Periaert; G. Fontaine et al.: "Survival of infant with arginosuccinic aciduria to three months of age." J. Pediatr., 86:639, 1975.

Shih, V.E. and M.L. Efron: Urea cycle disorders in Stanbury-Wyngaarden-Frederickson. The metabolic basis of inherited diseases. 3ed. McGraw-Hill, New York, 1972. p. 370.

ALMSTRÖM SYNDROME

Syndromography

Clinic

It begins in the newborn with hypotonia, to which is added obesity between 2-4 years, diffuse and not exaggerated. Height delay and moderate IQ.

Facies: small eyes, anti-mongolian palpebral fissure, narrow forehead and sometimes microcephaly. Hypogonadism: small penis and testicles, gynecomastia in adults.

Limbs: clinocampodactyly of the fifth finger and fork deformity of the fourth and fifth fingers with ulnar deviation of the hands, genu valgum and flat feet. These patients also present nystagmus, divergent strabismus, polar cataract, progressive decrease in vision and photophobia. The fundus shows retinitis pigmentosa. Nervous deafness, saccharine diabetes and primary gonadal insufficiency.

Paraclinical examinations

Blood. Normality of T4, TSH, HGH, LH, LSH and 17- -ketosteroids. Low testosterone values. Normal gonadotropins.

Karyotype. In order to confirm chromosomal sex and exclude structural defects, mainly of chromosome 15.

Syndromogenesis and etiology

The production mechanism is poorly understood, possible dysfunction of the pituitary gland, hypothalamus or higher regulatory centers. There are several conditions that affect a single person in the family with karyotype 46,XY which suggests a recessive inheritance linked to the X chromosome.

This autosomal recessive disorder is similar to Biedl-Bardet syndrome, which also has polydactyly and mental retardation absent in Almström.

Diabetes mellitus is the result of resistance to insulin action, but the molecular basis is not known.

The etiology is genetic.

Literature

Charles, S.J. et al.: “Almström's syndrome: further evidence and autosomal recessive inheritance and endocrinological dysfunction". J. Med. Genet.. 27:590-592, 1990.

Vásquez, L.B.; DL. Hurst and J.E. Sotos: "X-linked hypogonadism gynecomasia mental retardation short stature and obesity a syndrome" J. Pediat., 94:56, 1979.

CONGENITAL ALOPECIA SYNDROME

Synonymy

S. of universal congenital atrichia. S. of universal hereditary congenital baldness.

Syndromography

Clinic

Children are born without hair with cephalic baldness located over cranial sutures, but there is lanugo in the usual places, even eyebrows and eyelashes are missing.

There is a less severe form, which begins between 6 and 12 years of age, predominating in post-pubertal females.

Alopecia appears in a rounded shape with defined edges; it can grow until it merges with others and produces total alopecia areata.

Paraclinical examinations

Histological study of hair. These present morphology in the form of an exclamation point.

Syndromogenesis and etiology

It is proposed that the congenital form has a genetic etiology with autosomal dominant inheritance. The basic defect is not known. (See McKusick.)

ANDERSENI SYNDROME

Synonymy

S. of Andersen's triad. S. of pancreatic infantilism.

Cystic fibrosis of the pancreas.

Syndromography

1. Bronchiectasis.
2. Cystic fibrosis of the pancreas.
3. Vitamin A deficiency.

Syndromogenesis and etiology

Andersen I syndrome is part of the mucoviscidosis syndromes. The etiology is unknown. It is suspected to have a genetic basis.

Literature

Andersen, D.H.: "Cystic fibrosis of the pancreas and its relationship to celiac disease", Amer. J. Child., 56:344-99, 1938.

ANDERSEN II SYNDROME

Synonymy

S. of amylopectinosis. S. glycogenosis IV type Cori. S. of familial liver cirrhosis with abnormal glycogen storage. S. of 1,4-glucan-6-glucosyltransferase deficiency.

Syndromography

Clinic

This syndrome is very rare or difficult to diagnose. Only 3 cases have been reported, all in males.

It begins in childhood and all manifestations develop between 7 and 12 months after birth. There are repeated respiratory infections. The first symptoms are: anorexia, vomiting, hypotonia. The development is poor. The abdomen becomes prominent with progressive hepatomegaly and collateral circulation, the kidneys also increase in volume.

Shortly after, jaundice begins and progressively symptoms of liver cirrhosis (hematemesis, ascites, portal hypertension).

Physical examination. Marked hepatosplenomegaly, increased abdominal volume, ascites, jaundice, collateral circulation, hematemesis due to esophageal varices; sometimes large kidneys.

Paraclinical examinations

Blood. Moderate anemia. Decreased hyperglycemic response to adrenaline and glucagon. At the beginning, liver function tests are little altered but they deteriorate over the months and hyperbilirubinemia, hypoprothrombinemia and hypoproteinemia also occur.

Glucagon stimulation gives a flattened curve. The diagnosis is established by demonstrating a deficiency in the activity of the enzyme 1,4-glucan-6-glucosyltransferase in leukocytes or fibroblast culture. All cases die before 32 months with diarrhea, hematemesis and hyperpyrexia.

Liver biopsy. Advanced liver cirrhosis. Vacuolated cells containing an atypical glycogen that is stained blue by iodine. Low values or absence of 1,4-glucan-6- -glucosyltransferase. They are also found in the spleen, ganglia, lung, heart, and muscles.

Syndromogenesis and etiology

This syndrome is due to an accumulation in the liver and other organs of an atypical glycogen similar to plant amylopectin caused by a defect in the debranching enzyme, amyl (1,4 to 1,6), transglucosidase. As a consequence, glycogen accumulates, causing liver cirrhosis, which leads to the child's death.

Prenatal diagnosis is based on the levels of debranching enzyme in cultured amniotic fluid cells or chorion cells. The primary disorder is a defect in the debranching enzyme (1,4-1,6-amyl) transglucosidase.

The etiology is monogenic genetics. It is transmitted as an autosomal recessive inheritance.

Literature

Andersen, D.H.: "Studies on glycogen disease with report of a case in which the glycogen was abnormal." In Najjar, V.A.: Carbohydrate metabolism. Baltimore, Johns Hopkins, 1952. pp. 28-42.

Benson, P.T. and A.H. Fenson: Genetic biochemical disorders. Ith. ed. Oxford Med. Pub., New York, 1985. pp.349-351.

Brown, B.I. and H.D. Brown: "Branching enzyme activity of cultured aminocytes and chorionic villi: prenatan testing for type IV glycogen storage disease." Am. J. Hum. Genet., 44:378-381, 1989.

Howell, R.R.: In Stanbury-Wyngaarden-Frederickson: The metabolic basis of inherited disease. 3 ed. McGraw-Hill., New York, 1972, p. 149.

Mosses, S.W. and A. Gutman: "Inborn errors of glycogen metabolism." Adv. Pediat., 19:95, 1972.

Pearson, C.M.: "Glycogen metabolism and storage diseases of type II, IV and V." Amer. J. Clin. Path. 50:29-43, 1968.

ANENCEPHALIA SYNDROME

Synonymy

S. of acrania. S. of encephalorrhaphy. S. of exencephaly. S. sequence anencephaly.

Syndromography

Clinic

The defect is congenital, the cranial vault and almost the entire brain are missing, a translucent membrane replaces the scalp. The face looks large, grotesque, there is exophthalmia, a large nose, and poorly formed ears. Maro's reflection is partial, they do not cry, nor do they sneeze; They present periodic crises in flexion or extension.

Paraclinical examinations

Fetal ultrasound. This test is of interest for prenatal diagnosis, as well as the analysis in maternal serum and amniotic fluid of alpha feto protein whose concentrations from week 16 are abnormally high.

Syndromogenesis and etiology

Its etiology is multifactorial; the defect occurs in the normal closure of the anterior region of the neural tube, which should conclude after 28 days. This closure defect gives rise to a sequence of events: incomplete development of the brain, with degeneration, incomplete development of the skull and facial and auricular alterations.

Literature

Giroud, A.: Causes and morphogenesis of anencephaly. Ciba Foundation. Symposium on Congenital Malformations, pp. 199-218, 1960.

ANGELMAN SYNDROME

Synonymy

S. of happy doll. S. for happy puppet. S. of children's dolls.

Syndromography

Clinic

This syndrome occurs in childhood and is characterized by continuous head movements, with extension and flexion.

rhythmicity of head and hands. This movement can be stopped voluntarily and ceases during sleep and during intentional movements. The movements imitate those of a doll or puppet, continuous protrusion of the tongue. They present a generalized fine tremor, hypersensitivity to skin stimuli.

Peculiar facies with certain mandibular prognathism, the skull is small and brachycephalic. The crisis of unmotivated laughter without an emotional component is characteristic. They may have epilepsy, ataxic gait and in some patients pigmentary choroiditis.
Children have significant mental retardation with IQ50. The clinical picture is accompanied by marked hydrocephalus and obesity.

Physical examination. Brachycephaly, microcephaly, occipital flattening or depression, prognathism, incomplete development of the choroid.

Paraclinical examinations

Skull x-ray. Vertical inclination of the base.

Electroencephalogram. Typical pattern expressed by: 1, absence of hypsarrhythmia. 2,. presence of symmetrical and synchronous peak wave activity of 2 p/s.

Pneumoencephalogram.

Echoencephalography.

Study of cerebrospinal fluid.

Syndromogenesis and etiology

Very rare form of childhood epilepsy. A 15q 1lq 13 deletion has been reported in the chromosome of maternal origin; it receives the genetic material of that region of chromosome l5 only from its father, which suggests that a phenomenon of genomic imprinting due to uniparental disomy, as in Prader-Willi Syndrome, is involved in the etiology of this syndrome.

Literature

Angelman, H.: *Puppet children, a report on three cases". Develop. Med. Child. Neurol., 7:681-88, 1965.

Benton, J.W.; G. Nelhaus; P.R. Huttenlocher et al.: "The bubble-head syndrome: report of a unique truncal tremor associated with third ventricular cyst and hydrocephalus in children." Neurology, 16:725-299, 1966.

Berg, J.M. et al.: "Angelman's ('happy puppet') syndrome." Am. J. Dis. Child., 123:72., 1972.

Bower, B.D. and P.M. Jeavons: "The happy puppet' syndrome." Arch. Dis. Child., 42:298-302, 1967.

Hall, J.G.: "Genomic imprinting: review and relevance to human diseases", Am J. Hum, Genet., 46: 857-873. 1990.

Williams, C.A. and J.L. Frias: The Angelman's ('happy puppet') Syndrome". Am. J. Med. Genet., 11:453-455, 1982.

ANOPHTHALMIA SYNDROME

Synonymy

S. of true or primary anophthalmos.

Syndromography

It is always a bilateral malformation, without other malformations. Sometimes it is difficult to differentiate between extreme microphthalmia and cryptophthalmos.

Paraclinical examinations

Ocular ultrasound. To differentiate from microphthalmia and cryptophthalmos, where variable degrees of development of the optic cup are observed.

Syndromogenesis and etiology

Of genetic etiology, the parents of affected children are consanguineous, which confirms autosomal recessive inheritance. This anomaly is due to a failure in the formation of the optic cup and only the ectodermal structures are absent.

MASS ASPIRATION SYNDROME

Synonymy

S. of fetal aspiration. S. of neonatal atelectasis. S. of meconium aspiration.

Syndromography

Clinic

Aspiration of meconium-stained amniotic fluid can cause respiratory disorders in the newborn ranging from mild tachypnea to respiratory failure.

Generally it is a full-term newborn weighing more than 250 g. The severity of symptoms depends on the amount of meconium aspirated. If aspiration is minimal, tachypnea and slight cyanosis will present with a favorable evolution.

In massive aspiration the child may be intensely depressed at birth with irregular respirations, panting and intense cyanosis with scattered rales in both lung fields; he may die from hypoxemia and hypercapnia.

Sometimes there is rigidity and convulsions in more severe cases.

severe.

Physical examination. There is intercostal retraction, pale asphyxia, brain damage in severe cases, with decreased resonance to chest percussion, rales may or may not be heard and the temperature is normal if superinfection does not develop.

Paraclinical examinations

Radiology. It can be seen:

1. Increase in the anteroposterior diameter of the thorax.
2. Flattening of the diaphragms.
3. Irregular patches of infiltrate that radiate from the hila to the peripheral lung fields that are accompanied by areas of atelectasis.

4. Pneumothorax and pneumomediastinum.
5. Pleural effusion.

Pathology. Lungs are observed with an increase in their consistency, poorly aerated, bronchi filled with mucus or liquid, collapsed alveoli and others expanded. Elements of the amniotic fluid are recognized. Edema and bleeding areas.
There may be dilation of the right heart in some cases and hemorrhage and cerebral edema may coexist.

Syndromogenesis and etiology

It is proposed that intestinal hypoxia, physiological defecation or vagal stimulation as a result of compression of the umbilical cord can produce meconium elimination during fetal life. On the other hand, in a fetus that suffers intrauterine asphyxia, sudden inspiration or postnatal gasping may occur, which would be produced by aspiration of meconium fluid.

Literature

Farber, S. And J.L. Wilson: “Atelectasis of the newborn: a study and critical review." Amer. J. Dis. Child., 46:572-89, 1933.

Gooding, C.A. et al.: "Roentgenographic analysis of meconium aspiration the newborn." Radiology, 100;131, 1971.
Kachner, J. et al.: "La detress respiratoire du nouveaune par inhalation massive de liquide amniotique: Etude de 65 observations." Arch. Fr. Pediatr., 26:743, 1969.
Robert, S. and 0. Bacsik: "meconium aspiration syndrome North American Pediatric Clinics. Inter-American Ed. August 1977, pp. 467-478.

ASPLENIA SYNDROME

Synonymy

S. of Ivemark. S. of polysplenia.

Syndromography

Clinic

The congenital absence of hypoplastic spleen is accompanied by complex cardiac malformations, malposition and developmental defects of abdominal organs, and abnormal lobulation of the lungs.

Paraclinical examinations

Abdominal ultrasound. For diagnosis of the indicated malformations.

Echocardiogram

Peripheral blood smear. Heinzy Howell-Jolly bodies as hematological signs of absence of spleen.

Syndromogenesis and etiology

Genetic etiology is suggested, reports of affected siblings and consanguineous couples support autosomal recessive inheritance.

It is explained by a primary lateralization defect that in turn determines a defect in normal asymmetric development.

Literature

Arnold, G.L.: D. Bixler and D. Girod: *Probable autosomal recessive Inheritance of polysplenia, situs inversus and cardiac defects in an Amish family." Am. J. Med. Genet., 16:35-42, 1983.

CONGENITAL ATHIREOSIS SYNDROME

Synonymy

S. of athyreotic cretinism.

Syndromography

Clínic

The onset is prenatal, although they are born with high weight and height. Its frequency is 1 per 5,000 births and shows a 2:1 female predominance. It is characterized by prolonged jaundice, large fontanels, hypothermia and sometimes cyanosis. The syndrome establishes itself in the infant between 2-3 months. Features:
hypertelorism, eyelid bulging (myxedematous), flat nose with a wide and depressed root, mouth half-open through which the macroglossic tongue protrudes.

Low frontal hairline.

Delayed psychosomatic development, as well as bone maturation.

The legs are short, general hypotonia, bulging abdomen with umbilical hernia, square hands and short fingers. The skin It is pale, yellowish, cold, mottled with myxedema in the supraclavicular fossae. In addition, there is bradypsychia, hypothermia, constipation, and bradycardia. They seem too good, because they hardly cry and when they do the cry is hoarse and short.

Paraclinical examinations

Blood. T4 less than 6.25/100 mL in the neonate (0 to 5.25 in infants). TSH less than 20 micro U/ WL.

Radiology. Absence of epiphyseal, distofemoral and proximotibial nuclei in the neonate.

Thyroid functional study. Radioactive iodine uptake (zero in neonates) and less than 10% in infants. Scintigraphy:
absence or great thyroid hypoplasia, sometimes ectopic.

Iodine uptake. Low or absent.

Syndromogenesis and etiology

Congenital athyreosis syndrome is due to a deficiency of T4, essential for extrauterine somatic and brain growth. The etiology is genetic, most cases are sporadic although there is a familial recessive form (29%).

Most are dysgenesis of the gland, others are due to antithyroid substances in the mother and more rarely due to congenital infections (toxoplasmosis). There are frequently thyroid remnants at the base of the tongue.

Literature

Curling, T.B.: Med. Chir. Trans. London, 33:303, 1850 (cited by Andersen)

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PULMONARY ATRESIA SYNDROME WITH INTACT VENTRICULAR SEPTEM

Syndromography

Clinic

In most cases this anomaly is associated with a hypoplastic right ventricle, but with thick walls and lined with thick endocardium, the orifice of the tricuspid valve is small (type I). In 15-20% of cases the right ventricle is normal or large and the tricuspid valve is functionally incompetent (type I). The intermediate forms between both extremes are common.

Physical examination. Cyanosis is recognized from the first day of birth and heart failure is early, and cardiomegaly is common. The second heart sound is perceived isolated and systolic murmurs of maximum intensity in the middle part of the left sternal border are frequent. The patient presents an appearance of acute illness and distress from the first days of life. The differential diagnosis must be established with Ebstein's syndrome.

The treatment is surgical with high mortality during the operative act. The prognosis is bad.

Paraclinical examinations

Electrocardiogram. Tall, pointed P waves. If the right ventricle is small, signs of left ventricular hypertrophy are common and the QRS axis appears normal or deviated to the right. If the right ventricle is normal or large, right ventricular hypertrophy is noted.

Radiology. There is extreme variability in heart size, so it is generally normal in newborns with a small right ventricle, but it increases progressively during the first weeks of life. If the right ventricle is normal or large, there is marked cardiomegaly.

Cardiac catheterization. Demonstrates hypertension in the right atrium and ventricle.

Ventriculography. Shows the size of the ventricular cavity, atretic pulmonary valve, tricuspid regurgitation.

Syndromogenesis and etiology

The etiology is unknown

Literature

Cole, R.B.; A.J. Muster; M. Lev et al.: “Pulmonary atresia with intact ventricular septum.” Amer. J. Cardiol., 21:23-31, 1968.

Newfeld, E.A.; R.B. Cole and M.H. Paul: *Ebstein's malformation of the tricuspid valve in the neonate." Amer. J. Cardiol., 19:727- -731, 1967.

BEALS-HECHT SYNDROME

Synonymy

S. of Beals. S. of contracto-arachnodactyly.

Syndromography

Clinic

Long, thin limbs (dolichostenomelia) with arachnodactyly and ulnar deviation of the fingers. Contracture of the joints, especially knees, elbows and hands.
Kyphoscoliosis, short neck, varus metatarsals. Muscle hypoplasia. Auricular defect of the concha and the normal folds of the antihelix. Mitral valve prolapse.

The differential diagnosis is made primarily with Marfan syndrome.

Other malformations are micrognathia, coloboma of the iris, congenital malformations of the heart and aorta, subluxation of the patella. Contractural joint defects may improve, but scoliosis is progressive.

Paraclinical examinations

Radiology and echocardiogram. They confirm the manifestations already described.

Syndromogenesis and etiology

The etiology is genetic with autosomal dominant transmission.
nante. The basic defect is not known. It could be related to connective tissue alterations.

Literature

Beals. R.K andE. Hecht: "Delineation of another heritable disorder of connective tissue." J. Bone Joint. Surg. (Am), 53:987-990, 1971.

COLLODION BABY SYNDROME

Synonymy

S. of lamellar exfoliation of the newborn. S. de Carini. Seeligman S. S. of the baby crocodile.

Syndromography

This syndrome is a rare congenital condition in which the child is born completely covered by a constrictive membrane. similar to parchment or collodion. This constrictive membrane that immobilizes the child causes eyelid ectropion. Within 24 hours of birth, a fissuring or exfoliation process begins that allows large keratinized plaques to be eliminated, which coincides with a great improvement. Some cases recover completely. The injuries can be partial and/or affect the entire body. The large quadrilateral scales can be so thick that they look like armor plates.

They usually suffer complications: sepsis and loss of proteins and electrolytes.

Syndromogenesis and etiology

The etiology is unknown. Some authors consider it to be a clinical form of lamellar ichthyosis (see).

Literature

Carini, A.: “Di una forma attenuata della considetta ittiosi sebacea.” Gior. Italian. Bad. Vener. 36:82-88, 1895.

Dentz. C.L. and J. Altman: “Lamellar ichthyosis, the natural clinical cause of collodian babies." Arch. Derm. (Chicago), 97:3-13, 1968.

Seeligman, E.: De Epidermis, Imprimis neonatorum desquqmqtione. Inaugural dissertation, Berlin, 1841.

BECKWITH-WIEDEMANN SYNDROME

Synonymy

S. de Wiedemann-Beckwith. S. of exomphalo-macroglossia-gigantism. S. of hypoglycemia with macroglossia.

Syndromography

Clinic

Craniofacial dysmorphia due to prominent eyes with relative infraorbital hypoplasia, nevus flammens. Capillary on forehead and eyelids, metopic ridge, large fontanels, prominent occiput, malocclusion with tendency to mandibular prognathism. Characteristic linear folds on the earlobes. From a clinical point of view, this syndrome is characterized by:
macrosomia with large muscle masses and thick subcutaneous tissue, macroglossia and omphalocele. Large kidneys with renal medullary dysplasia, pancreatic hyperplasia with excess islets, fetal adrenocortical cytomegaly, which is a consistent fact.
Interstitial cellular hyperplasia of the gonads and amphophilic hyperplasia of the pituitary.

Other malformations: posterior diaphragmatic eventration, cardiomegaly, hepatomegaly, slight microcephaly, hemihypertrophy, Wilms tumor, hepatoblastoma, clitorimegaly, large ovaries, hyperplastic and bicornuate uterus, cardiac hamartoma.
Immunodeficiency. Moderate, mild mental retardation or normal intelligence.

The natural history of the disease can begin with hydramnios and a high incidence of prematurity, the average birth weight is 4 kg and the length is 52.6 cm. There is difficulty in neonatal adaptation, apnea, cyanosis and seizures may occur, the large tongue occludes the respiratory tract and makes feeding difficult. Hypoglycemia responds to treatments with hydrocortisone analogues and requires only 1 to 4 months.

Paraclinical examinations

Radiology. Accelerated bone maturation, widened and deflected metaphyses with overconstriction of the diaphysis.

Abdominal ultrasound. In search of visceromegaly. Alpha-feto-serum protein. Every 6 months until 6 years of age for the diagnosis of Wilms tumor and hepatoblastoma.

Blood glucose and hematological studies. Neonatal pancytopenia,

Syndromogenesis and etiology

Partial duplications of the short arms of chromosome 11 have been reported.

The etiology is unknown, generally cases are sporadic.

Literature

Beckwith, J.B.: "Extreme cytomegaly of the adrenal cortex, omphalocele, hyperplasia of kidneys and pancreas, Leydigszell hyperplasia: another syndrome?" Gather. Soc. Amer. Res. (Los Angeles, Cal.), 1963.

Beckwith, J.B.; C.I. Wang; G.N. Donneell, et al.: "Hyperplastic fetal visceromegaly with macroglossia omphalocele, cytomegaly of adrenal fetal cortex, postnatal somatic gigantism and other abnormalities: Newly recognized syndrome." Proc. Amer. Pediat. Soc., Seattle, Washington, June, 16-18, 1964 (abst. No. 41).

Beckwith, J.B.: “Macroglossia, omphalocele, adrenal cytomegaly, gigantism and hyperplastic visceromegaly.” Birth Defects, 5:188- -190, 1969.

Maziri, M. et al.: “Abnormality of chromosome 11 in patients with features of Beckwith-Wiedemann syndrome". J. Pediatr., 102:873-975, 1983.

BERARDINELLI SYNDROME

Synonymy

S. of Seip. S. of total lipodystrophy with acromegaloid gigantism. S. of congenital progressive lipodystrophy. S. of total lipodystrophy. S. of lipoatrophic diabetes. S. of generalized lipodystrophy. S. of lipodystrophy-gigantism. S. of infantile acromegaloid gigantism. S. of congenital lipodystrophic diabetes with acanthosis nigricans. S. of hypertrophic muscular lipodystrophy.

(See cerebral gigantism syndromes in children.)

Syndromography

Clinic

It is characterized by complete loss of adipose tissue, generalized hyperpigmentation, and hypertrichosis that may be present at birth. It can be seen in both sexes, it predominates in the female sex (2:1). It is associated with mental retardation and symptoms of diabetes.

Physical examination. Increased height, generalized loss of fat, with the appearance of premature aging are observed in children. Emaciated skin, acanthosis nigricans in the armpits, groin, hands and neck. Large hands and feet, muscle hypertrophy. Hypertrichosis with abundant, curly and coarse hair. Genitals with an increase in size, in the male the penis and in the female an increase in the clitoris without other signs of virilization.

Umbilical hernia, there may be hepatosplenomegaly and cardiomegaly that is not constant. Marked subcutaneous veins. Corneal opacity and advanced dentition. Acromegaloid features, macrocephaly and macroclitoris without sexual precocity. In adults, a normal or slightly elevated height is observed.

Exams p paraclinical s

Blood. Hyperglycemia, neutral fat hyperlipemia with marked elevation of triglycerides.

Hyperproteinemia. Normal pituitary, gonadal and adrenal hormones.

Ophthalmological examination. Corneal lipodystrophy.

Basal metabolism. Increased.

Radiology. In children, an advanced bone age is demonstrated in relation to the chronological one: 1, absence of fat in the soft tissues that results in a low homogeneous density; 2, increased bone density;3, widening of the cortex of tubular bones; 4, prominent and hypertrophic epiphyses; 5, adolescence of skeletal maturation; 6,

dense transverse bands on the vertebrae.

Urine. Albuminuria, cylindruria.

Pneumoencephalography. In some cases ventricular dilation and sometimes cerebral gigantism.

Liver biopsy. Fatty infiltration, round cell infiltration, glycogen deposits.

Syndromogenesis and etiology

Possibly it is a genetically determined disorder (affected hormones and frequency of consanguinity of the parents have been seen).

The possibility of a homozygous autosomal recessive state is raised; it may not be a single clinical entity.

It has many elements in common with cerebral gigantism syndrome in children and Russell syndrome. Different genetic defects can cause the syndrome. It may manifest at birth or present a later onset.

Disorders in the secretion of fat mobilizing agents and hypopituitary or hypothalamic pathology have been reported.

An antagonistic action of insulin and mobilization of fats has been detected in urine. Possible dysencephalic syndrome similar to leprechandism as an anti-Prader-Willi. It constitutes a rare form of thesaurismosis with pituitary hypereosinophilism. The basic defect is at the level of the insulin receptor. There appear to be at least five gene mutations: 1, damage to receptor biosynthesis; 2, receiver transportation defect; 3, decreased insulin binding affinity; 4, damage to tyrosine kinase activity; 5, accelerated degradation of the receptor.

Literature

Berardinelli, W.: Undiagnosed endocrine metabolic syndrome: report of two cases". J. Clin. Endocrin. Metab., 14:195, 1954.

Fairney, A., et al.: "Total lipodystrophy." Arch. Dis. Child., 44:368, 1969.

Lawrence, R.D.: Lipodystrophy and hepatomegaly with diabetes, lipaemia, and other metabolic disturbances; a case throwing new light on the action of insulin". Lancet, 1:724, 773, 1946.

Milunsky, A., V.A. Cowie and E.C. Donoghue: "Cerebral gigantism in childhood, a report of two cases and a review of the literature." Pediatrics, 40:395-402, 1967.

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Seip, M.: "Lipodystrophy and gigantism with associated endocrine manifestations: a new diencephalic syndrome?" Paediatr Act. 48:555-74, 1959.

Taylor, S.I., et al.: "Genetic basis of endocrine disease 1: molecular genetics of insulin resistant diabetes mellitus." J. Clin. Endocr. Metab. 73:1158-1163, 1991.

BERENDES-BRIDGES-GOOD SYNDROME

Synonymy

Familial chronic granulomatosis S. Chronic granulomatous disease of childhood.

Syndromography

Clinic

This syndrome predominates in men (80%). There are repeated bacterial and fungal infections, mainly in the first 2 years of age. It is rare for carriers to suffer serious infections. The infected areas are those that are most exposed, such as the perinasal and perioral areas. Liver abscesses (staphylococcal) and osteomyelitis of the bones of the hands, feet and limbs are common.

Physical examination. Hepatosplenomegaly, lymphadenopathy. Soft tissue abscesses, purulent lymphadenitis, conjunctivitis, diarrhea and facial rashes are also observed.

Paraclinical examinations

Blood. Anemia, leukocytosis with neutrophilia. Erythrosedimentation - elevated. Hypergammaglobulinemia. Intermediate defects of the function of neutrophils, distinguishing two central populations lular when incubated with nitroblue tetrazolium.

Bacteriological examination of the material obtained. Gram-positive germs (Staphylococcus aureus). Gram-negative germs (Serratia marcescens, Klebsiella and Pseudomonas).

Radiology. Pulmonary infiltrates.

Syndromogenesis and etiology

Defective microbicidal capacity of phagocytic cells and abnormal reaction of oxidative metabolism during phagocytosis. In most males there is an X-linked recessive inheritance pattern. In most females the pattern appears to be autosomal recessive.

Unaffected parents and siblings are normal.

Literature

Bachner, R.L. and D.G. Nathan: Leukocyte oxidase." Science, 155:835-836, 1967.

Berendes, H.; R.A. Bridges and R.A. Good: *A fatal granulomatosis of childhood, the clinical study of a new syndrome." Minnesota Med., 40:309-312, 1957.

BIBER-HAAB-DIMMER SYNDROME

Synonymy

Reticular type corneal dystrophy. Biber-Haab-Dimmer degeneration. Buckler III.

Syndromography

Clinic

It affects males and females and begins in late adolescence or youth (20-30 years of age), with progressive loss of vision.

Physical examination. Presence of grayish, transparent corneal lines (like cotton threads) that mainly occupy the area between the center and the corneal periphery (without reaching the sclerocorneal limbus).

Paraclinical examinations

Corneal biopsy. Hyaline degeneration of the lamellar stroma without mucopolysaccharide deposition.

Syndromogenesis and etiology

This syndrome is a monogenic defect of autosomal dominant inheritance.

The etiology is genetic.

Literature

Biber, H.: "Ueber einige seltenere Hornhauterkrankungen. Inaugural Sissertation." Zurich, 1890 (Cited by Haab, O.: Die gittrige Keratitis. Ztschr. E Augenh. 2:235-246, 1899).

Feigin, R.D. and D.B. Caplan: "Corneal opacities in infancy and childhood." J. Pediat., 69:383-392, 1966.

BIELSCHOWSKY SYNDROME

Synonymy

S. de Jansky-Bielschowsky.

Syndromography

Clinic

This syndrome begins between 3 and 4 years of age. It is characterized by amaurotic atrophy of the optic nerve, extrapyramidal muscle hypertonia, idiocy, dysphagia, and marasmus. Unlike Tay-Sach syndrome, the cherry red spot next to the macula is not present.

Syndromogenesis and etiology

It is cerebroretinomacular degeneration with brain atrophy and macular degeneration of the retina. The white matter of the spinal cord and the brain can undergo fascicular demyelination. This entity is considered to be infantile neuronal ceroid lipofuscinosis for which there are several genes.

The etiology is genetic with autosomal recessive inheritance.

Literature

Bielschowsky, M.: “Ueber spatinfantile familiare amaurotische Idiotie mit Leinhirnsymptomen.” Deutsch. Z. Nervenheilk., 50:7-29, 1914.

Jarvela, L: "Infantile neuronal ceroid lipofuscinosis (CLN 1): linkage dysequilibrium in the Finnish population and evidence that variant late infantile form (variant CLN 2) represents a non-allelic locus." Genomics, 10:333-337, 1991.

BLACKFAN-DIAMOND SYNDROME

Synonymy

St. Josephs-Diamond-Blackfan. S. de Kaznelson. S. of erythrogenesis imperfecta. S. of idiopathic erythroblastopenia. S. of congenital hypoplastic anemia. S. of primary red cell aplasia.

Syndromography

Clínic

Symptoms occur at birth or within the first 6 months. There is growth retardation. Defects in sexual maturation. Asthenia. Shortness of breath. Low birth weight.

Pallor. There is no jaundice. Presence of small congenital anomalies. Occasionally there is hepatomegaly, splenomegaly is rare and begins to decrease after repeated transfusions.

Paraclinical examinations

Blood. Normocyticnormochronic or hyperchromic anemia. Reticulocytes below 1%. Platelets and leukocytes normal.

Medulogram. Hypoplasia of the erythropoietic system.

Syndromogenesis and etiology

An abnormality of tryptophan metabolism, a congenital abnormality of sensitivity to erythropoietin that causes a functional deficiency of erythroid precursors, and more recently, alterations of hematopoietic growth factors and ligands for the tyrosine kinase receptor have been suggested.

The heterogeneity in pathogenesis and the observation of autosomal recessive and dominant inheritance suggest genetic heterogeneity.

Literature

Abkowitz, J.L. et al.: "Diamond-Blackfan anemia: in vtro response of erythroid progenitor to the ligand for c-kit. Blood, 78:2198-2202, 1991.

Diamond, L.K. and K. D. Blackfan: *Hypoplastic anemia". Amer. J. Dis. Child., 56:464-67, 1938.

Tartaglia, A.P.; S. Propp; AP Amarose, et al.: "Chromosome abnormality and hypocalcemia in congenital erythroid hypoplasia (Blackfan-Diamond's syndrome)." Amer. J. Med., 41:990-99, 1966.

BLEPHAROPHIMOSIS SYNDROME

Synonymy

S. of familial blepharophimosis.

Syndromography

Clínic

This syndrome is characterized by a facial dysmorphic pattern in which short palpebral fissures with displacement of the internal canthus, depression of the nasal bridge, palpabral ptosis, and strabismus stand out. Incomplete development of the ear pavilions. Two types are described:

Type I. Presents variable hypogonadism with infertility in women.

Type II. There is no fertility defect. Patients may occasionally have mental retardation and heart defects.

Paraclinical examinations

Karyotype. For the study of hypogonadism, in some cases structural chromosomal aberrations with breakpoints in 3q21-23 or 3q2 deletion have been reported.

Syndromogenesis and etiology

It is a syndrome of genetic etiology with autosomal dominant inheritance and with different characteristics for each type.

Type I. With infertility that is inherited as a sex-limited autosomal dominant trait, the causes of infertility are unknown. Men transmit the trait to both sexes but women do not reproduce.

Type II. Autosomal dominant inheritance without sex differences.

The chromosomal alterations detected in some patients with the syndrome have suggested the location of the gene in 3q2.

Literature

Alvarado, M.; M. Bocian and A.P. Walker: "Insterstitial deletion of the long arm of chromosome 3: case report, review and definition of a phenotype". Am. J. Med. Genet., 27:781-786, 1987.

BÖRJESON-FORSSMAN-LEHMANN SYNDROME

Synonymy

S. of mental retardation-epilepsy-endocrine disorders.

Syndromography

Clinic

Almost all cases are men; some female carriers with moderate mental retardation.

Physical examination. Facies: thick due to diffuse obesity with narrow palpebral slits and large, non-deformed ears. Trunk: gynecomastia due to obesity and abundant abdominal adipose panniculus. Genu valgum limbs. Central nervous system: intense mental retardation with IQ between 20-40.
lifelong epilepsy. Growth is deformed with final height between 136 and 156 cm.

Paraclinical examinations

Urine. 17-ketosteroids and low gonadotropins.

Functional study. Radioactive iodine uptake, protein iodine and amino acid chromatography, normal.

Syndromogenesis and etiology

The reported families allow us to propose recessive inheritance linked to the X chromosome.

Linkage has been demonstrated that allows us to assume the location of the gene in Xq26-27.

The etiology appears to be genetic.

Literature

Baar, H.S. and J. Galindo: The Börjeson-Forssman-Lehmann syndrome". J. Ment. Def. Res., 9:125, 1965.

Börjeson, M.; H. Forssman and D. Lehmann: "Combination of idiocy, epilepsy, hypogonadism, dwarfism, hypometabolism and morphologic peculiarities in inherited as an X-linked recessive syndrome." Proc. 2nd, Int. Cong. Ment. Delay Vienna (1961) Part. 1:188, 1963.

Holmes, L.B. et al.: Mental retardation. MacMillan C.. New York, 1972, p.25.

Turner, G.; A. Gedeon, J. Mulley et al.: "Börjeson-Forssman-Lehmann syndrome: Clinical manifestations and gene localization to Xq26-27". Am. J. Genet. 34:463-469, 1989.

BYLER SYNDROME

Synonymy

S. Fatal. S. familiar. S. of intrahepatic cholestasis. Progressive cholestatic familial cirrhosis.

Syndromography

Clinic

Both sexes are affected in family members of Jews who bear the surname Byler. It begins in the first years of childhood. It is characterized by recurrent episodes of jaundice accompanied by infectious processes. Epistaxis. Pruritus.
Diarrhea with foul, loose stools, followed by jaundice.

Delayed growth in weight and height

Physical examination. Icterus, protruding abdomen. Appearance of a sick person. Hepatosplenomegaly. The prognosis is poor, they die between 20 months and 8 years of age.

Paraclinical examinations

Blood. Hyperbilirubinemia. Elevated alkaline phosphatase.

Normal or low cholesterol. Hyprothrombinemia

Stool. Light in color with steatorrhea.

Urine. Dark due to bilirubinuria.

Laparoscopy. It is essential to exclude bile duct atresia.

Syndromogenesis and etiology

Congenital biochemical abnormality with recurrent cholestasis due to a defect in excretion of conjugated bile salts and formation of lithocholic acid that damages cells.

It is transmitted by autosomal recessive inheritance.

Literature

Clayton, F.J., FL. Iber; B.H. Bucbner, et al.: "Byler's disease: fatal familial intrahepatic cholestasis in an Amish kindred (abstracted)", J. Pediat., 67: 1025-28, 1965.

Clayton, F.J.; FL. Iber, B.H. Bucbner et al.: "Byler's disease: fatal familial intrahepatic cholestasis in an Amish kindred." Amer. J. Dis. Child., 117:112, 24, 1969.

DANCING WRIST HEAD SYNDROME

Synonymy

In Anglo-Saxon literature Bubble-head doll syndrome.

Syndromography

Clinic

It occurs in children aged 5-10 years, somewhat thick and with a large head. It is characterized by continuous head movements, with rhythmic extension and flexion of the head and hands at a rate of 2-3 movements per second, in the form of denial or affirmation that is sometimes transmitted to the trunk, but can be inhibited at will and subsides during sleep.

In addition, a generalized fine tremor and hypersensitivity to cutaneous stimuli may be observed. Mental retardation, marked hydrocephalus and obesity.

Physical examination. Minimal patellar and Achilles hyperreflexia, occasional mild ataxia of the trunk or limbs. Normal intelligence.

The head circumference is around 60 cm.

Endocrine symptoms: absence of thirst, sexual precocity, amenorrhea and tendency to adipose-genital habit.

Paraclinical examinations

Pneumoencephalography and ventriculography. Increase in finger prints.

Echoencephalography

Study of cerebrospinal fluid.

Syndromogenesis and etiology

This syndrome occurs after the age of 5 and is a consequence of a cyst in the third ventricle or hydrocephalus due to occlusion of the Sylvian aqueduct.

Literature

Benton, J.W.: G. Nelhaus; P.R., Huttenlocher et al.: "The bubble-head syndrome: Report of a unique truncal tremor associated with third ventricular cyst and hydrocephalus in children." Neurology, 16:725-29, 1966.

Nelhaus, G.: "The bubble-head doll syndrome: a 'tic' with a neuropathologic basis." Pediatrics, 40:250. 1967.

Ruisman, D.S.; S.H. Tucker and L. Schut: *Slow tremor and macrocephaly. Expanded version of the bubble-head-doll syndrome". J. Pediat., 87:63, 1975.

CANDIDIASIS-ENDOCRINOPATHY SYNDROME

Syndromography

Clinic

Onset during infancy with extensive mucocutaneous moniliasis infection evident soon after birth. Hypoparathyroidism is common and acute adrenal insufficiency may occur. Diabetes is rare.

Paraclinical examinations

Immunological study. From the beginning, organospecific antibodies against a variety of endocrine glands can be discovered. It can be associated with: pernicious anemia, sprue, chronic active hepatitis and membranoproliferative glomerulonephritis.

Syndromogenesis and etiology

There is defective cellular immunity against Candida albicans; some patients show more generalized allergy.

A cause-and-effect relationship between monilia infection and endocrinopathy has not been established. The disorder has occurred in siblings, sometimes as a result of consanguineous unions, and the disease can be inherited in an autosomal recessive manner. No relationship with the HLA system has been demonstrated, but affected individuals may suffer from immunoglobulin A deficiency and hypergammaglobulinemia.

The function of suppressor T cells may be deficient, but the immune profile may vary even between siblings.

FACE-NECK SYNDROME

Synonymy

S. for phenothiazine.

Syndromography

Clinic

The diagnosis is made by observing the following clinical examination: the tongue is deformed and protruding, spasm of the chewing muscles, speech disorders, pharyngeal constriction, dysphagia and trismus are confirmed; Spasmodic torticollis frequently exists, it is always related to the history of ingestion of preparations that contain phenothiazine such as: Trilafon, Decentan, Melleril, Siquil, Vesprin,

etc.

Syndromogenesis and etiology

It is due to phenothiazine poisoning.

Literature

Robinson, A.S.: *Neck phase syndrome related to Phenothiazine drugs." JAMA 173(5):504-506, 1960.

ELF FACE SYNDROME

Synonymy

S. of hypercalcemia, supravalvular aortic stenosis and elf face. S. of hypocalcemic face. S. Williams. S. of the elf- of gnome. S. of Donohue.

Syndromography

Clinic

It is present at birth and is characterized by mental retardation, delayed bone development, and sexual precocity.

Possibly it occurs exclusively in females. There are some cases reported in men that are denied by other authors. In females, puberty is precocious. Low birth weight. Facies: elfin face with widened nasal bridge, prominent jaws, large and asymmetrical lips with prominence of the upper one, pointed palate, convergent strabismus, overlapping of the teeth of the lower arch, large ears.

Physical examination. Hirsutism. Characteristic facies, hypertherorism, long ears. Hypertrophy of nipples and external genitalia. Decreased height and weight for age. Reduced adipose panniculus, large hands and feet. Acanthosis nigricans and cryptorchidism. Systolic murmur, hypertension, hoarse voice. Hallux varus, clinodactyly of the little fingers, sunken nails, hunched chest.

Paraclinical examinations

Blood. Hypoglycemia. Low alkaline phosphatase. Hypercalcemia, possible sphingomyelin abnormality.

Radiology. Increased calcification in the skull, long bones and vertebrae. Evidence of supravalvular aortic stenosis, pulmonary stenosis, or septal defects.

Urine. Low excretion of gonadotropins, increased excretion of 17-ketosteroids.

Syndromogenesis and etiology

Family condition, possible recessive inheritance, parental consanguinity and absence of chromosomal anomaly. Pathogenic mechanism: intrauterine follicular maturation. Excess vitamin D has also been raised in the early fetal period, because it has been reproduced in rabbits.

The etiology is unknown.

Literature

Donohue, W.L.: "Dysendocrinism." J. Pediatr., 32:739-48, 1948.

Dupont, A. and J. Clausen: "The elfin facies syndrome." Lancet, 1:209, 1968.

Friedman, W.F. and F.M. Lorer: "The relationship and aortic stenosis syndrome." Pediatrics, 43:12, 1969.

Jone, K.L. and D.W. Smith: "The Williams elfin face syndrome: A new perspective." J. Pediatr., 86:718, 1975.

Langdon-Down, J.L.H.: Observations on an ethnic classification of idiots clinical lectures and report. London, 1866.

Rogers, D.R.: "Leprechaunism (Donohue's syndrome)." Amer. J. Clin. Path., 45:614-19, 1966.

Rowe, R.D. and R.E. Cooke: “Vitamin D and craniofacial and dental anomalies of supravalvular stenosis.” Pediatrics, 43:1, 1969.

"CAT EYE SYNDROME"

Synonymy

S. coloboma of the iris. S. of anal atresia. S. de Trip (22) (pter-qll).

Syndromography

Clinic

There is no incidence data, but the reported cases do not exceed 50.

Dysmorphology: the most relevant clinical manifestations are: coloboma of the iris, which can also be accompanied by coloboma of the choroid and retina, as well as antimongoloid deviation of the palpebral fissures and hypertelorism.

Other malformations: anal atresia with rectovestibular fistula.

Congenital heart diseases including anomalies of pulmonary venous return and persistence of the left superior vena cava.

Se han reportado casos con agenesia renal.

Growth is normal in most patients.

Neurology: moderate mental deficiency or normal intelligence with some degree of emotional retardation.

Paraclinical examinations

Ophthalmological exams.

Echocardiogram.

Kidney ultrasound.

Karyotype.

Syndromogenesis and etiology

Presence of a chromosomal imbalance due to an extra marker chromosome recognized as derived from the identical segments of chromosome 22 persisting from the satellites, the short arms of the centromere and up to the 22q11 segment, which gives rise to a quadrupled segment 22.

Literature

Shinzel, A. et al.: "The Cat Eye Syndrome': Dicentric small marker chromosome probably derived from a No. 22 (tetrasomy 22 pter ql1) associated with a characteristic phenotype. Report of 11 patients and delineation of the clinical picture". Hum. Genet, 57:148-158, 1981.

BRAIN-COSTOMANDIBULAR SYNDROME

Syndromography

Clinic

There is growth deficiency of prenatal onset. The facies is characterized by severe micrognathia with glossoptosis, a sunken short palate and soft palate. El tórax es pequeño de forma acampanada. Mental deficiency in surviving patients, hearing loss. Occasionally, microcephaly and clinodactyly of the fifth finger and redundant skin can be observed. 40% die in the first year and half of these in the first month due to respiratory failure.

Paraclinical examinations

Radiology. In the thorax, an interruption between several or all of the ribs of 0.5-2 cm is observed between the ossified costal arch and the cartilaginous costal portion, especially from the fourth to the tenth ribs. Anomalies of costal insertion to the vertebra.

Abnormal tracheal cartilaginous rings, elbow hypoplasia.

Syndromogenesis and etiology

It is a gene mutation but its inheritance is not well delineated; In some families it behaves as autosomal recessive and in others as autosomal dominant.

Literature

Leroy, J. G. et al.: "Cerebro-costo-mandibular syndrome with autosomal dominant inheritance." J. Pediatr., 99:441-444, 1981.
Smith, K.G. and K.C. Sekar: “Brain-costo-mandibular syndrome.” Clin. Pediatr.. 24:223, 1985.

CRYPTOGENETIC CYANOSIS SYNDROME IN CHILDHOOD

Syndromography

Clinic

It is observed in one-month-old newborns who, in perfect health, suddenly become cyanotic, remaining alert and active; or with a slight decrease in activity and signs of irritability and lack of appetite.

Physical examination. Pallor and cyanosis, tachycardia, there are no respiratory alterations and the liver is of normal size. There is smooth and firm splenomegaly without lymphadenopathy and slight elevation of rectal temperature.

Paraclinical examinations

Blood. Leukocytosis with neutrophilia. Spectrophotometric hemoglobin tests have not been performed.

Syndromogenesis and etiology

It is possible that it is an infection with the formation of methemoglobin. It is curious to note that the manifestations disappear in 1-3 weeks in the following order: first cyanosis, then splenomegaly and finally tachycardia.

The etiology is unknown.

Literature

Wilkinson, S.J.: “Acute cryptogenic cyanosis in early infancy.” Illinois Med. J.. 91:29-35, 1947.

CLARKE-HADFIELD SYNDROME

Synonymy

S. of congenital hypoplasia of the exocrine pancreas.

Syndromography

Clínic

This syndrome affects both sexes and begins in youth.

Poor growth and development, poor musculature, lack of subcutaneous fat. Digestive disorders similar to mucoviscidosis syndrome (see). Usually absence of respiratory complications. Sometimes it is associated with hematological alterations (see S. de Shwachman).

Paraclinical examinations

Stool. Abundant, fat.

Pancreatic tests. They are useful in blood, urine and feces.

Sweat test. Normal.

Pancreatic biopsy. There is no risk of pancreatic fistula since there are no ferments.

Literature

bodian, M.; W. Sheldon and R. Lightwood: "Congenital hypoplasia of the exocrine pancreas." Paediat Act. (Stockholm), 53:282-93, 1964.

Clarke, C.; and G. Hadfield: "Congenital pancreatic disease with infantilism." Quart. J. Med., 17:358-64, 1924.

COCKAYNE SYNDROME

Synonymy

S. of leukodystrophy-microcephaly-striocerebellious calcification. S. of retinal atrophy-dwarfism. S. of trisomy 10. S. of progeroid dwarfism. S. de Neill-Dingwall. S. of pseudoprogeria.

Syndromography

Clinic

This syndrome appears in the second year of life and is characterized by skin photosensitivity with pigmentation and scars. The dermatitis is similar to that of lupus erythematosus with cutaneous photosensitivity, erythema, loss of subcutaneous fat, hyperpigmentation and atrophy of the face. The ears are large and protrude resembling the face of Mickey Mouse. Senile facies, wrinkled face with loss of fat, sunken eyes, thin nose, prognathism.

The limbs are disproportionately long with large hands and feet and flexion contractures. Kyphosis. There is decreased vision, optic atrophy with retinal pigmentation and cataracts. The pupillary response to mydriatics is poor.

Hypohidrosis and decreased tearing. Pendular nystagmus.

There are intracranial calcifications and it is accompanied by defective physical and mental development and mental retardation.

Progressive hearing loss, nerve deafness and other anomalies.

There is spastic weakness and ataxia of the limbs. On occasions it may be accompanied by athetosis, amyotrophy with abolition of tendon reflexes and somewhat slowed nerve conduction. In some cases there is hepatomegaly.

Motor lesions and cerebellar alterations lead to death before the age of 30.

Paraclinical examinations

Chromosome study Skin biopsy

Radiology. Thickening of the cranial shell. Intracranial calcifications. Relative shortening of the phalanges and metacarpus; thickening of the epiphysis of the phalanges ("eburnized epiphysis").

Blood. Hyperbetaglobulinemia. Renal acidosis. Decreased endogenous creatinine clearance.

Audiometry and electroretinogram

Syndromogenesis and etiology

There are at least three types of Cockayne syndrome, all of which are transmitted by autosomal recessive inheritance. The genetic defect consists of the alteration of the enzymes necessary to repair damage induced in DNA by ultraviolet light, which as is known affects the transcription of active DNA; This explains the pleiotropic effect of the gene in relation to neurodegeneration.

Cancer is not a characteristic of this syndrome nor are infectious complications common. It is unknown

exact mechanism of this disorder, although a metabolic origin has possibly been invoked. Calcifications of the basal nuclei of the telencephalon and cerebellar have been observed, as well as leukodystrophy similar to that which occurs in Pelizaeus-Merzbacher syndrome.

It is inherited as an autosomal recessive trait with 100% penetrance.

The etiology is genetic.

Literature

Cockayne, E.A.: "Dwarfism with retinal atrophy and deafness." Arch. Dis. Child.,11:1-8, 1936.
Foley, J.: "Calcification of the corpus stratum and dentate nuclei occurring in a family." J. Neurol. Neurosurg. Psychiatry., 14:253, 1951.
Moossy, J.: "The neuropathology of Cockayne's syndrome." J. Neuropath. Exp. Neurol., 26:654-60, 1967.
Nance, M.A. and S.A. Berry: "Cockayne's syndrome: review of 140 cases." Am. J. Med. Genet. 42:68-84, 1992.
Paddison, R.M. et al.: "Cockayne's syndrome" Derm. Int. 2:195- -202 (Oct.-Dec.), 1963.
Venega, J. et al.: The genetic defect in Cockayne's syndrome is associated with a defect in repair of U.V.-induced DNA damage in transcriptionally active DNA". Proc. Nat. Acad. Sci.,87:4707- -4771, 1990.

COFFIN SYNDROME

Synonymy

S. de Coffin-Lowry.

Syndromography

Clínica

Mild to moderate growth deficiency of postnatal onset; severe mental deficiency; relative muscle weakness.

Coarse facies with maxillary hypoplasia, hypertelorism.

Wide nose with wings and thick nasal septa. Thick lips with a prominent and everted lower lip and prominent ears.

Hypodontia, malocclusion and separated teeth. Pectus carinatun. Large hands with accessory transverse hypothenar ridge.

Ligamentous hypermobility, flat feet.

Paraclinical examinations

Radiology. Short and bifid sternum, vertebral defects with thoracolumbar scoliosis. Alterations in the bone development of the epiphyses. Short terminal phalanges in a drumstick shape.

Syndromogenesis and etiology

It is transmitted by X-linked inheritance in which homozygous males develop the syndrome and female carriers may have some clinical manifestations. The basic defect is unknown.

Literature

Hunter, A.G.S., M.W. Partington, and J.A. Evans: "The Coffin-Lowry' s syndrome. Experience from four centers." Clin. Genet., 21:321-323, 1982.

COFFIN-SIRIS SYNDROME

Syndromography

Clinic

Moderate to mild growth deficiency with prenatal onset. Severe mental retardation, hypotonia, microcephaly, facies coarse with thick lips. Generalized hirsutism with a tendency to have poor and scattered hair. Hypoplasia or absence of the nail of the fifth finger and foot with hypoplasia of the nails of the rest of the fingers and toes. Joint laxity with radial dislocation of the elbow. Gastrointestinal anomalies, obstruction secondary to redundant gastric mucosa. Other anomalies include shortening of the forearm, sunken palate, Dandy-Walker anomaly, hypoplasia or partial agenesis of the corpus callosum, and other brain anomalies.

Recurrent respiratory tract infections.

Paraclinical examinations

Radiology. Coxa valga, small patellas. Delayed bone age.

Brain ultrasound and computerized axial tomography. To specify the malformations described.

Therotropic hormone. Normal. It is indicated for the differential diagnosis with congenital hypothyroidism.

Syndromogenesis and etiology

Not well clarified yet.

Bibliography

Coffin, G.S. and E. Siris: Mental retardation with absent fifth fingernail and terminal phalanx." Am. J. Dis. Child., 119:433- -435, 1970.

PYRIDOXINE-DEPENDENT SEIZURE SYNDROME

Synonymy

S. of pyridoxine-dependent encephalopathy.

Syndromography

Clinic

It begins within a few hours or days of life; some cases begin in the womb at 5-7 months, others a few hours after birth. It is characterized by hyperirritability and hyperacusis (the knocks of the incubator where the child is located trigger rhythmic automatisms in this child). Bad development.

Intense, brief, repeated tonic-clonic seizures that do not respond to usual anticonvulsants and improve with the administration of high doses of vitamin B6. They may present nystagmus.

There is a history of the mother receiving high doses of pyridoxine during pregnancy. The convulsions are preceded by fainting with paleness and rotating movements of the eyes. Treatment with pyridoxine produces a dramatic resolution of seizures and with maintenance treatment recurrences are avoided.

The prognosis is fatal without treatment; With it, seizure episodes are controlled, but mental retardation persists.

Paraclinical examinations

Electroencephalogram. There is marked dysrhythmia and slow wave activity with high potential, with a hypsoarrhythmic appearance.

Findings that are present when there are signs of incipient seizures. After pyridoxine injection, a decrease in slow wave activity appears.

Cerebrospinal fluid. Hyperproteinorrachia.

Syndromogenesis and etiology

It is due to an inborn error of metabolism, with increased needs for pyridoxine, permanently: this "dependency" is characterized by three features: genetic etiology, precise pharmacological dose of pyridoxine and biochemical specificity of the disorder.

It is transmitted in an autosomal recessive manner, affecting both sexes.

The etiology is genetic. The basic defect is due to insufficiency of the enzyme glutamic acid decarboxylase in the conversion of glutamic acid into alpha aminobutyric acid (GABA), the major inhibitory neurotransmitter of the CNS.

Literature

Hunt, A.D.; J.R. Stokes; W.W. Crory et al.: Pyridoxin dependency: report of a case of intractable seizures in an infant". Pediatrics, 13:140-145, 1954.

Scriver, C.R.: "Vitamin B6-dependency and infantile seizures." J. Pediat., 26:62, 1960.

Waldinger, C.: "Pyridoxine deficiency and pyridoxine-dependency in infants and children." Postgrad. Med., 35:415, 1964.

HYPOPLASTIC LEFT HEART SYNDROME

Synonymy

S. of hypoplasia of the left heart.

Syndromography

Clínic

It begins in the first to second week of life with congestive heart failure (it is the only heart disease capable of causing this condition in the neonate). It is characterized by severe dyspnea, progressive due to failure of the left cavities. Progressive cyanosis, of low intensity, which has a tiger-skin appearance and is accentuated in the terminal phase.

Physical examination. Imperceptible pulse, even in the carotid.

Blood pressure: the same in arms and legs. Thickened heart with slight left parasternal bulging, single second tone, systolic ejection click at left border and apex;

In half of the cases, a murmur of intensity 2-6, systolic, located in the upper left parasternal region. The gallop rhythm is frequent. It represents the main cause of mortality due to heart disease in the first week of life.

Paraclinical examinations

Radiology. Cardiomegaly with bulging of the right border and increased pulmonary vascularity.

Electrocardiogram. Right ventricular hypertrophy and deviation to the right of the electrical axis, increased R waves in the right precordial and beaked P waves.

Angiocardiography. Hypoplasia of the ascending aorta and rapid filling of the descending aorta from the right ventricle.

Syndromogenesis and etiology

The fundamental mechanism that causes the syndrome is the absence of function of the left ventricle, which obstructs the return circulation from the lungs and the mixed blood in the right ventricle passes, in part, to the pulmonary artery and part through the ductus (which persists) to the ascending aorta: this leads to heart failure.

It is transmitted in an autosomal recessive manner.

The etiology is genetic.

Literature

Lev, M.: "Pathologic anatomy and interrelationship of hypoplasia of the aortic tract complexes." Lab. Invest., 1:61, 1952 (cit. P. Noonan).

Noonan, J.A. and A.S. Nadas: "The hypoplastic left heart syndrome." Pediat. Clin. N. Amer., 5:1029, 1958.

Quero, M.; L. Pérez Díaz and J. Bourgon Codina: Congenital heart disease: Diagnosis and treatment. Scientific Ed. Medical. Madrid 1973, p. 20.

CHILDREN'S CHORIOCARCINOMA SYNDROME

Syndromography

Clinic

It occurs in both sexes. It appears between 5 weeks and 7 months, manifesting itself as anemia crisis with hemoptysis, hematemesis, hematuria and/or melena.

Physical examination. There is hepatomegaly, breast enlargement, pubic hair growth. A palpable abdominal mass may appear.

Paraclinical examinations

Chest radiology. It shows the presence of a tumor mass in the lung. Urine. There is marked elevation of chorionic gonadotropin.

Pathology. Choriocarcinoma can be demonstrated in the lungs, liver, subcutaneous cellular tissue, adrenal glands and brain.

Syndromogenesis and etiology

It is a metastatic choriocarcinoma from the placenta acquired during intrauterine life. The prognosis is unfavorable but its diagnosis allows treatment to be established for the mother.

Literature

Witzleben, C.L.: and G. Bruninga: "Infantile choriocarcinoma: a characteristic syndrome." J. Pediat.. 73:374-378, 1968.

CRIDU CHAT SYNDROME

Synonymy

S. of monosomy 5p. S. of the cat's meow.

Syndromography

Clínic

Estimated incidence of 1 in 50,000 live births. Sexual relationship 7F: 5M.

It is characterized by abnormal crying similar to the meowing of a cat due to its high-pitched and tense tone.

Dysmorphology: microcephaly, round, moon-shaped face, hypertelorism, epicanthus, wide and flat nasal bridge. Poorly defined jaw angle. Full cheeks. With age, although microcephaly persists, the face lengthens, there is poor implantation of the teeth, and micrognathia is more evident. At birth they have a special cry that is reminiscent of a cat's meow, possibly due to hypoplasia of the larynx.

Other malformations: diffuse cerebral atrophy, ventricular dilation, cerebral atrophy, hydrocephalus. Optic atrophy, strabismus.

Congenital heart disease.

Neurology: hypotonia with poor reflexes, hypertonia of the limbs and spastic gait appear with age. Severe mental retardation, no language development. They can reach adults.

Paraclinical examinations

Karyotype.

Electroencephalogram. It is usually abnormal.

Bone x-ray. Scoliosis, flat feet, rib and vertebral anomalies.

Ultrasound of the brain and other organs.

Computerized axial tomography.

Syndromogenesis and etiology

Chromosomal imbalance due to deletion of the short arms of chromosome 5. Most of the characteristics described are observed even with the loss of a small segment 5p14p15.

Literature

Berg, J.M. et al.: "The cri-du-chat syndrome in adolescents and adults." J. Pediatr., 77: 782,1970.

Lejeune, J. et al.: "Trois cas de délétion partielle du bras cour du chromosome 5", C. R. Academic Sci. [D]Paris).

CRIGLER-NAJJAR SYNDROME

Synonymy

S. of familial non-hemolytic jaundice with kernicterus. S. of Rotor-Crigler-Najar. S. of congenital hyperbilirubinemia.

Syndromography

Clinic

This condition exists in two forms. Type I is the severe one from a clinical point of view (it is the one originally described by Crigler and Najjar) and is caused by a total lack of glucuronyltransferase. Type II is more moderate, since there is only a partial deficiency of the enzyme.

Type I. It is rare. It begins in the infant on the first day and persists thereafter and is accompanied by unconjugated hyperbilirubinemia of about 20 mg/dL, sometimes higher. Kernicterus occurs several weeks later, due to the accumulation of unconjugated bilirubin in the gray nuclei at the base of the brain.

Absence of conjugated bilirubin in bile that loses its color. There is no response to phenobarbital. Liver tests are normal, as is liver histology. Phototherapy somewhat, albeit temporarily, reduces unconjugated bilirubin. Affected children die in the first weeks or months, others in the first year of life. On rare occasions they have survived into the second to third decades. It corresponds to that originally described by Crigler and Najjar.

Type II. As glucuronyltransferase deficiency is partial, the clinical picture is less severe. Jaundice may not appear until adolescence or may be moderate. Neurological complications of the kernicterus type are very rare.

Paraclinical examinations

Blood. Hyperbilirubinemia is 6-20 mg/dL, mainly based on monoconjugates. Delayed clearance of bilirubin in the blood.

Liver biopsy. Normal.

Syndromogenesis and etiology

Phenobarbital is effective as an enzyme inducer and a rapid drop in blood bilirubin is observed.

The condition is relatively benign.

Inheritance is dominant with variable expressivity in type I. Recessive in type II.

Literature

Crigler, J.F. and V, Najar: *A congenital familial non-hemolytic jaundice with kernicterus". Pediatrics, 10:169-79, 1952.

Schmidt, D.: Hyperbilirubinemia. In Stanbury, J.B. and D.S. Frederickson: The metabolic basis of inherited disease. McGraw-Hill, New York, 1966.

CROSS SYNDROME

Synonymy

S. Oculocerebral-hypopigmentation.

Syndromography

Clinic

It was first discovered in a consanguineous Amish family with 4 affected children.

It is manifested from birth by ocular anomalies and albino appearance of skin and hair. The skin is almost depigmented, although it lacks the total whiteness of albinism; creamy-white hair; some pigmentary nevi.

Eyes: microphthalmia with cloudy, vascularized microcorneas, gross nystagmus and total blindness; spastic ectropion.

Central nervous system: Around 3 months of age, athetoid movements of the fingers and toes appear, with progressive spasticity and strong mental retardation;

hypertoniae hyperreflexia without reaching clonus. When school age arrives, contractures of the shoulders, elbows, hips and knees develop, and sucking and grasping reflexes persist. Profound mental retardation and progressive disability.

Hypogonadism: in males cryptorchidism; In females, when they reach age, secondary sexual characteristics do not appear. Mouth: gingival fibromatosis.

Paraclinical examinations

Karyotype, amino acids and electroencephalogram. Nor-evils.

Syndromogenesis and etiology

There is a great reduction of melanocytes in the hair bulb, which are barely impregnated with tyrosine in vitro.

It is transmitted in an autosomal recessive manner. Frequently the parents of these patients are consanguineous. The few known cases are of Amish origin (Ohio), published by Cross, McKusick and Breer.

The etiology is genetic.

Literature

Cross, H.E.; GOES. McKusick and Q. Breer: “A new oculocerebral syndrome with hypopigmentation.” J. Pediat., 70:398, 1967.

Go R.M. and R.J. Gorlin: Atlas of the Face in Genetic Disorders. Ed. Mosby, 1977.

CURTIUS I SYNDROME

Synonymy

S. of congenital unilateral gigantism. S. of hemimacromrosomia.

Syndromography

Clínica

It may present with marked hypertrophy of a part of the body or face or be limited to a joint or the alveolar arch and teeth. There is hemihypertrophy of the face.

Sometimes limited.

There may be syndactyly, nail or skin dystrophy. Sometimes microcephaly; dental enamel isplasia with hypodontia.

It is generally accompanied by endocrine disorders:

hypogenitalism, breast hypoplasia and sometimes psychological alterations.

Syndromogenesis and etiology

It is considered a probably hereditary condition.

Literature

Curtius, E: “Kongenilater Partieller Riesenwuchs mit Endocrinen Storungen.” Deut. Arch. Klin. Med., 147:310-19, 1925.

Moortgat, P.: Syndromes a noms propres. Paris, Pretat, 1966.

ALPHA-1-ANTYTRIPSIN DEFICIT SYNDROME

Syndromography

Clínica

The clinical manifestations of this syndrome are fundamentally given by neonatal intrahepatic cholestasis, which constitutes 5-10% of cholestatic icters in the newborn. In older children and adults it causes pulmonary emphysema and is associated with other conditions such as glomerulonephritis, pancreatitis and joint diseases.

Physical examination. In the most common form of presentation.

Jaundice, hepatomegaly, intermittent hypocolia, dark urine and early splenomegaly are observed.

The prognosis of these children is variable. Sometimes the lesion progresses to liver cirrhosis, with portal hypertension and cirosis, leading to early death in the first decade of life. In other cases the cholestatic manifestations improve and the progression towards cirrhosis is slow. Finally, there are patients who achieve greater survival and reach adulthood although they present variable degrees of liver dysfunction.

Paraclinical examinations

Blood. There is a pattern of cholestasis characterized by elevation of bilirubin with predominance of the direct fraction, increase in alkaline phosphatase, which can reach up to 4 times its normal value, and moderate increase in aminotransferases.

Protein electrophoresis shows a decrease or absence of alpha-1-globulins.

Pi system phenotype. The Pizz phenotype is pathological.

Determination of the antitryptic power of serum. Diminished.

Abdominal ultrasound. He shows signs of portal hypertension with hepatosplenomegaly.

Laparoscopy. In the neonatal period it shows a cholestatic liver. Subsequently, elevations and irregularities appear on the liver surface, taking on the appearance of a mixed macronodular and micronodular cirrhosis.

Liver biopsy. Presents inflammatory infiltrate of the portal spaces with increase in connective tissue and necrosis of the hepatocytes. These contain a substance that is revealed by the PAS stain (periodic acid Schiff), which is resistant to the action of diastase. It has been suggested that this substance may be made up of non-secreted alpha-1-antitrypsin molecules.

Syndromogenesis and etiology

This syndrome is genetic and is transmitted in an autosomal recessive manner, regulated by a system of multiple autosomal codominant alleles that are located on the long arm of chromosome 14. These elements regulate the production of alpha-1-antitrypsin (the most important of the plasma alpha-1-globulins) and inhibit multiple proteolytic enzymes, including trypsin, which decreases in a range between 10 and 15% of the normal figure in patients with the disease. (pizz phenotype).

Literature

Alagille, D. and M. Odievre: "Diseases of the liver and bile ducts in children." Medical Scientific Editorial, Cap. 3 pp. 33- -68. Barcelona, 1981.

Fragoso, T. et al.: *Value of determining the Pi system phenotype in alpha-1 antitrypsin deficiency." Rev. Cub. Ped., 58(3):3 15-321, 1986.

TRYPSYNOGEN DEFICIT SYNDROME

Synonymy

Trypsinogen deficiency disease.

Syndromography

Clinic

It begins shortly after birth, with intermittent vomiting, formed stools and little weight gain that remains stationary despite eating with appetite. Its frequency is one case per 10,000 births. It is characterized by edema, pallor, hypochromotrichia. All symptoms return with replacement treatment.

Physical examination. Edema from 8 weeks with evident godet. Anemia. Hypochromotrichia with partial hair lightening, as occurs in kwashiorkor.

Paraclinical examinations

Blood. Normocytic normochromic anemia with slight macrocytosis (does not respond to medication). Moderate reticulocytosis, severe neutropenia, marked hypoproteinemia of all fractions.

Sweat tests. Negatives.

Urine. Normal.

Duodenal tube. Incomplete absence of trypsinogen.

Stool. Negative tryptic reaction, with steatorrhea (fats constitute 50% of the dry weight). Proteinorrhea, with loss of 60% of ingested nitrogen.

Syndromogenesis and etiology

In this syndrome there is no ability to synthesize pancreatic trypsinogen, which is why chymotrypsin and carboxypeptidase are also low, since these enzymes are

formed from the corresponding proenzymes, by trypsin.

Trypsinogen deficiency deprives pancreatic juice of its proteolytic capacity, which causes protein malabsorption and malnutrition which, in turn, facilitates steatorrhea and accentuates malnutrition.

This syndrome has a genetic etiology and is transmitted in an autosomal recessive manner.

Literature

Gryboski, J.: Gastrointestinal problems in the infant. Ed. Saunders, Philadelphia, 1975, p. 460.

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Townes, P.L.: M. Bryson and G. Miller: "Further observation on trypsinogen disease: report of a second case." J. Pediat., 71:220, 1967.

Townes, P.L.: "Proteolitic and lipolytic deficiency of the exocrine pancreas." J. Pediat., 75:221, 1969.

Townes, P.L.; M. E. Bryson and G. Miller: "Further observation trysinogen deficiency disease; report of a second case." J. Pediat., 71:220-24, 1967.

DI GEORGE SYNDROME

Synonymy

S. of the third and fourth pharyngeal pouches. S. of congenital absence of thymus and parathyroid. S. of thymic aplasia.

Syndromography

Clinic

It affects both sexes, early onset in the first year of life.

Severe and frequent infections, particularly respiratory infections due to various microorganisms. Diarrhea, hypocalcemic tetany in the newborn. Facial findings: wide nose, retracted upper lip, hypoplastic jaw, low insertion of the ears, fusion of the helix and antehelix.

Mental retardation. Serious infections (fungi, viruses, pyogens).

Physical examination. Tetany, convulsions. Chvostek and Trousseau signs. Pathological fissures of the lips, nose and ears; micrognathia; congenital heart disease (especially right aortic arch and tetralogy of Fallot). Dehydration, lung and mucosa infection. Severe malformations: microcephaly, heart disease, etc.

Paraclinical examinations

Blood. Anemia, progressive lymphopenia, hypocalcemia, hyperphosphoremia.

Radiology. Absence of thymic shadow in the first week of birth, anomalies of the aortic arch, esophageal atresia, tracheoesophageal fistula, congenital heart

Immunological tests. Poor response to phytohemoglutinin, delayed immunity deficit, rejection of homoinfarction, normal serum immunoglobulin content, good antibody response, normal neutrophil function.

Syndromogenesis and etiology

It can be caused by: congenital absence, hypoplasia or ectopia of the parathyroid; absence of the thymus. These alterations produce the clinical picture.

It is due to embryopathy in the eighth week. Failure in the development of the third and fourth pharyngeal pouches, which gives rise to a malformation sequence in 60% of cases, is accompanied by a varied group of malformations.

Etiologically it has been associated with prenatal alcohol exposure and cases with 22 g deletion have been reported.

The etiology is genetic and most cases are sporadic; Only one familial case has been reported in siblings.

Literature

DiGeorge, A.M.: M.D. Cooper; R.D.A. Peterson and R.A. Good: "New concept of cellular basis of immunity." J. Pediat., 67:907- -08, 1965.

Jones, K.L.: Smith's recognizable patterns of humane malformation. 4th edition, 1988, W.B. Sanders Company, Philadelphia.

Lischner, H.W.; C.W. Wright and Le Riche et al.: "Partial DiGeorge syndrome with substantial cell-mediated immunity." Am. J. Dis. Child., 130:316, 1976

Lobdell, D.H.: "Congenital absence of parathyroid glands." Arch. Path., 67:412-15, 1959.

Taitz, L.S.; C. Zarate-Salvador and R.S. Schartz: "Congenital abscence of the parathyroid and thymus glands in an infant (I and IV pharyngeal poch syndrome)." Pediatrics, 38:412, 1960.

HEREDITARY NEPHROGENIC DIABETES SYNDROME

Synonymy

S. of the polyuro-polydipsic baby. (See ADH-resistant diabetes syndrome.)

Syndromography

Clinic

It manifests itself in male children of carrier women (who present only a minor degree of the same disorder). There is polydipsia, polyuria, resistance to ADH since childhood, dehydration, mental retardation.

Paraclinical examinations

Urine. Low density, inability to concentrate. Inability to respond to vasopressin.

Syndromogenesis and etiology

Sex-linked hereditary syndrome that causes serious disorders in men.

Literature

Carter, C. and M. Simpkiss: "Carrier state in nephrogenic diabetes insipidus. " Lancet, 2:1069-73, 1956.

Forssman, H.: "On hereditary diabetes insipidus, with special regard to a sex-linked form." Acta Med. Scand. Suppl., 159:1-196, 1945.

Williams, R.H. and C. Henry: "Nephrogenic diabetes insipidus transmitted by females and appearing during infancy in males." Ann. Int. Med., 27:84-95, 1947.

TRANSIENT DIABETES SYNDROME IN CHILDHOOD

Synonymy

S. of temporary congenital diabetes mellitus.

Syndromography

Clinic

This syndrome manifests itself in generally full-term, emaciated, low-weight newborns with an absence of subcutaneous fat and a senile appearance. It usually begins before 6 weeks of age. There is polyphagia, polyuria, dehydration, fever and progressive weight loss despite a correct diet.

The development of infections and abscesses is common. The differentiation of the state of transient diabetes and true diabetes mellitus can only be established when a sufficient period has elapsed for it to disappear or show its permanent nature. Treatment is with insulin (if blood glucose levels are greater than 200 mg%), antibiotics for infections and adequate diet and parenteral hydration if necessary.

The evolution is towards the correction of the metabolic defect between 7 days and 18 months when children present normal blood glucose and glucose tolerance curve.

Death may occur from starvation or marasmus.

Mental retardation may be observed in some cases.

Paraclinical examinations

Blood. Fasting hyperglycemia. Glucose tolerance curve similar to that of a diabetic.

Urine. Glucosuria.

Syndromogenesis and etiology

There are hypoplastic pancreatic islets incapable of producing adequate insulin. The mother has low blood glucose levels (dietary, overproduction of insulin) or placental disease that prevents normal transport of glucose from mother to child or a primary delay in the maturation of pancreatic islet cells.

Literature

Dacou Voutetakis, C. et al.: "Macroglossia transient neonatal diabetes mellitus and intrauterine growth failure. A new distinct entity." Pediatric, 55:127-13 1, 1975.

CAUDAL DYSPLASIA SYNDROME

Synonymy

S. of caudal regression. Caudal dysplasia sequence S.

Syndromography

Clinic

The structural defects of the caudal region in this malformation pattern are characterized by incomplete development of the sacrum that extends to a lesser degree to the lumbar vertebrae, absence of the body of the sacrum, flattening of the buttocks, shortening of the intergluteal cleft, dimpling of the buttocks, disruption of the spinal cord allowing secondary neurological damage that causes everything from incontinence of urine and feces to total neurological loss and Extreme loss of growth in the caudal region, reducing leg movement. The degree of severity of these clinical manifestations is variable, the most affected infants have flexion and abduction of the hips, popliteal membranes secondary to loss of movement.

Other common deformities are equinovarus foot and calcaneovalgus. There may also be renal agenesis, imperforate anus, cleft palate, microcephaly and meningomyelocele.

Paraclinical examinations

Radiology. Lumbosacral spine, pelvis and lower limbs allow bone alterations to be checked.

Kidney ultrasound.

Syndromogenesis and etiology

This malformation syndrome was grouped with sirenomelia, thinking that this represented its most severe form. Currently they are considered two unrelated entities. Sirenomelia and its associated defects are due to a vascular alteration of the vitelline artery, while caudal dysplasia is probably of heterogeneous etiology in which there seems to be a relationship with diabetic mothers.

Literature

Stewart, J.M. and S. Sotoll: "Familial caudal anomalous regression and maternal diabetes." J. Med. Genet., 16:17-20, 1979.

CRANIOMETAPHYSAL DISPLAY SYNDROME

Syndromography

Clinic

Increased thickness of the nasal bones and supraorbital area with hypertelorism and a relatively small nose. Variable proptosis of the eyes. Headaches and nasal narrowing with rhinitis. The disease becomes evident from childhood and patients can present serious problems with compression of the brain and cranial nerves. They may present facial paralysis, visual loss and deafness.

Paraclinical examinations

Radiology. Increased density of the skull base of the facial bones, jaw and cranial vault.
Variable absence of pneumatization. Slight to moderate metaphysal widening with diaphysal sclerosis.

Syndromogenesis and etiology

Of monogenic etiology, the type of inheritance is not well clarified. The basic defect seems to be related to bone morphogenesis with a defect in bone resorption and remodeling.

Literature

Gorlin, R.J.; J., Spranger and M. Koszalka: "Genetic craniotubular bone dysplasias and hyperostosis. A critical analysis". Birth Defects, 5: 79, 1969.
Penchaszadeh, V.B.; E.R. Gutiérrez and P., Figuero: “Autosomal recessive craniometaphyseal dysplasia". Am. J. Med. Genet., 5:43-45, 1980.

SPONDYLOMETAPHYSAL DYSPLASIA SYNDROME

Synonymy

S. of Kozlowski spondylometaphysal chondrodysplasia.

Syndromography

Clinic

This syndrome is characterized by growth deficiency, especially of the trunk, which begins between the first and fourth years of age, adult height ranges between 4 feet 3 inches and 5 feet. There is a short neck and trunk with dorsal kyphosis, pectus carinatum. Wandering gait with limitation of joint mobility and degenerative joint changes.

Sparaclinical examinations

Radiology. At the level of the spine in AP and L views, platyspondyly with anterior narrowing in the lumbar region is observed. Odontoid hypoplasia. In the pelvis the iliac wings are square and there is irregular flattening of the acetabulum. Irregular metaphyses, especially of the proximal femur with a short femoral neck. Hypoplastic carpal bones with delayed ossification.

Syndromogenesis and etiology

The etiology is genetic, autosomal dominant and also recessive inheritances are observed. There is a possibility of fresh mutations. The basic defect is not known.

Literature

Riggs, W. Jr. and R.L. Summitt: "Spondylometaphyseal dysplasia (Kozlowski) . Report of affected mother and son." Radiology. 101:375-377, 1971.

SCHMID TYPE METAPHYSAL DYSPLASIA SYNDROME

Syndromography

Clinic

Moderate low stature, 130-160 cm in adults. Bowing of the legs, with a waddling gait in the second year of age. Leg pain in schoolchildren. Moderate limitation in the complete extension of the fingers.

Paraclinical examinations

Radiology. Short long bones with widening and irregularity of the metaphysis. Arched tibias. Coxa vara and genuvarum. The epiphyses remain preserved.

Syndromogenesis and etiology

The etiology is monogenic genetics with autosomal dominant inheritance and variable expressivity. Although the basic defect is unknown, histological study reveals cartilage hypoplasia.

Literature

Rosenblom, A.L. and D.W. Schmid: “The natural history of metaphyseal dysostosis." J. Pediatr., 6:857-859, 1965.

JANSEN TYPE0 METAPHYSAL DYSPLASIA SYNDROME

Synonymy

S. of metaphysal dysostosis.

Syndromography

Clinic

Severe low stature with postnatal onset, in adults the height is around 125 cm. Unlike the other types of metaphysal dysplasias, it presents a facial dysmorphic pattern that is characterized by: a small face with prominent eyes, with frontonasal and supraorbital hyperplasia in adults and micrognathia. The patients have a small chest, flexion deformities of the joints especially knees and hips, waddling gait, short fingers, deafness and also present hypercalcemia.

Paraclinical examinations

Radiology (bone survey). Skull: hyperostosis with density of the skull base. Pelvis and metaphysis with areas that present cyst-like images due to loss of ossification.
cation.

Blood. Serum calcium.

Syndromogenesis and etiology

Of genetic etiology, new mutations arise in most cases and autosomal dominant inheritance. Although there is no knowledge of the basic defect, it is known that there are radiographic changes with age.

Literature

Charrow, J. and A.K. Poznaski: "The Jansen type of metaphyseal chondrodysplasia: confirmation of dominant inheritance and review of radiographic manifestations the newborn and adult." Am. J. Med. Genet., 18:321-325, 1984.

McKUSICK TYPE METAPHYSAL DYSPLASIA SYNDROME

Synonymy

S. of cartilage-hair hypoplasia.

Syndromography

Clinic

Low size (adult 107-147 cm). Short limbs with arched legs. Short hands, hypoplastic fingernails and toenails. Incomplete extension of the elbows. Fine, brittle, scattered hair. Joint hypermobility of hands and feet. Sometimes it is accompanied by a decreased cellular immune response and malabsorption in childhood.

Paraclinical examinations

Radiology. Long bones with irregular metaphyses. The tibia is shorter than the fibula.

Syndromogenesis and etiology

Of genetic etiology, monogenic of autosomal recessive inheritance. The basic defect is not known, however the cartilage cells are decreased, forming poorly organized columns.

Literature

McKusick, V.A. et al.: "Dwarfism in the Amish II. Cartilage-hair hypoplasia." Bull. Hopkins Hosp., 116:285-287, 1965.

Lux, S.E. et al.: "Chronic neutropenia and abnormal cellular immunity in cartilage-hair hypoplasia", New. Engl. J. Med., 282:23 1-234, 1970.

THORACIC DYSPLASIA SYNDROME

SUFFOCATING

Synonymy

S. of Jeune's thoracic dystrophy.

Syndromography

Clinic

Low stature from prenatal onset, small rib cage with pulmonary hypoplasia possibly secondary to the small chest, which causes death at an early age. Renal alterations such as tubular sclerosis and/or tubular cystic dysplasia. Liver disorders with variable proliferation of ducts.

Paraclinical examinations

Radiology (bone survey). Short costal arches with irregularity in the costochondral joint. Relatively short long bones with irregularity in the metaphysis and epiphysis, especially in the hands. Ulna and fibula short. Cone-shaped epiphysis and early fusion between epiphysis and metaphysis of the distal and middle phalanges. Early ossification of the head of the femur. Pelvis with hypoplasia of the iliac wings, horizontal acetabulum with projections towards the lower margins of the sacrociatic notch.

Kidney ultrasound.

Syndromogenesis and etiology

Of genetic etiology, it is transmitted with an autosomal recessive inheritance pattern. The basic defect is unknown; autopsies have shown disordered alterations in costochondral growth with hyperplastic proliferative cartilage and poor progress of endochondral mineralization. Fetal ultrasound at week 18 of gestation allows prenatal diagnosis.

Literature

Elejalde, B.R. et al.: "Prenatal diagnosis of Jeune's syndrome." Ame. J. Med. Genet., 21:433-435, 1985.

Okerklaid, F. et al.: “Asphyxiating thoracic dystrophy." Arch. Dis. Child., 52:758-760, 1977.

Shah, K.J.: “Renal lesions in Jeune's syndrome.” Br. J. Radiol. 53:432-434, 1980.

RESPIRATORY DISTRESS SYNDROME THAT SIMULATES CONGENITAL HEART DISEASE

Syndromography

Clinic

Term newborn with normal weight. He has extreme tachypnea without great retraction and respiratory moaning.

The lung sound is harsh and noisy. There is considerable enlargement of the heart; Progressive hepatomegaly, edema and cyanosis. The treatment is based on oxygen, digitalization and diuretics. The prognosis is good with high survival and good development contrary to other forms of respiratory distress.

Paraclinical examinations

Blood. 0, low arterial, there are only slight anomalies in the pH and PCO values.

Electrocardiogram and echocardiogram. To exclude major congenital heart disease.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Roberton, N.R.C.: "Severe respiratory distress syndrome mimicking cyanotic heart-disease in term babies Lancet, 2:1108-1110, 1967.

DOWN SYNDROME

Synonymy

S. of trisomy 21. "Mongolian idiocy" and "Mongolism" are terms criticized and not used particularly for their racial implications.

Syndromography

Clinic

Incidence: 1 in 1,000, increases exponentially with maternal age, 1 in 300 at age 35, 1 in 45 at 40 years, 1 in 15 at 45 years, 1 in 12.5 after 45 years.

Sex ratio 3M:2F.

Dysmorphology. The patients present a well-defined phenotype, a small skull with a flat occiput, a round face with a flat profile, oblique upward palpebral slits, the inner angle of the eye covered by epicanthus, false hypertelorism, short and scarce eyelashes, strabismus, blepharitis; Brushfield spots are observed in the irises when they are blue. The nasal bridge is flat, the nose small with anteverted nostrils.

Small mouth, thick, fissured lips, thick, protruding tongue, half-open mouth, there may be exfoliative glossitis, small, round ears with a prominent helix root, small ear canals, otitis media are common. Short, wide neck with excess skin at the nape of the neck. Globulous abdomen with diastasis recti anterior and umbilical hernia.

Short, wide hands and feet. In the hands there is brachymesophalangy, especially of the thumb and the fifth finger, a single palmar transverse fold is characteristic. On the feet there is a separation between the first and second toes with a deep groove between them. They present ligamentous hypermobility and muscle hypotonia. The pelvis is small with a decreased acetabular angle.

Other malformations: more than 45% of patients present congenital heart disease, the most frequent being atrioventricular canal, ventricular septal defect and atrial septal defect, persistence of the ductus arteriosus. There are digestive malformations such as duodenal stenosis, annular pancreas, megacolon, anal atresia and rectal prolapse. The average height in the adult male is 154 cm and 144 in the female.

Facial characteristics change with age. The onset of puberty is normal in both sexes. Mental retardation is constant and varies depending on age (IQ 50-70).

Syndromogenesis and etiology

Unbalanced chromosomal aberration involving chromosome 21. 92. 5% are free trisomies due to non-disjunction, possibly more frequent in maternal meiosis and depending on age.

Translocations due to centromeric fusion D/Go G/G, in 4.8% and mosaicisms in 2.7%.

Literature

Gadner, R.J. and G.R. Sutherland: Chromosome abnormalities and genetic counseling. Oxford University Press, New York, 1989.

Smith, D.W. and A.C. Wilson: The child with down's syndrome. Philadelphia, W. B. Saunders Co., 1973.

DYGGVE-MELCHIOR-CLAUSEN SYNDROME

Syndromography

Clínica

Low height at the expense of the trunk that begins postnatally appears around 18 months, the adult height is 128 cm or less. Mental deficiency with microcephaly. Protrusion of the sternum with barrel chest. Limitation of joint mobility. Wandering gait, genus valgus. Scoliosis, kyphosis, lordosis.

Paraclinical examinations

Radiology (bone survey). Flattened vertebrae with pointed anterior surface of the body and ossification defects such as notches, odontoid hypoplasia, iliac wings with irregularly calcified ridges. Lateral displacement of the epiphyses of the femoral head. Long bones with irregular metaphysises and epiphyses. Short metacarpal bones, particularly the first, short phalanges. Small carpal bones, conical epiphyses.

Syndromogenesis and etiology

Of genetic etiology, it has autosomal recessive inheritance. The basic defect is unknown.

Literature

Naffah, J.: "The Dyggve-Melchior-Clausen syndrome." Am.J. Hum. Genet., 28:607, 1976.

Spranger, J.; P. Maroteaux and V.M. der Kaloustian: "The Dyggve-Melchior-Clausen syndrome". Radiology, 114:415, 1975.Pri-ford

EEC SYNDROME

Synonymy

S. of ectodermal dysplasia, ectrodactyly and cleft lip and palate, EEC: from the English ectrodactyly -ectodermal dysplasia- -clefting.

Syndromography

Clínica

It presents alterations typical of ectodermal dysplasias:

thin skin with slight hyperkeratosis, hypoplastic nipples, little fine and light hair, hypoplastic nails, partial anodontia, microdontia and caries, photophobia, blepharophimosis, lacrimonasal duct defects, clear irises, blepharitis, dacryocystitis. Cleft lip with cleft palate, maxillary and molar hypoplasia.

Syndactyly, ectrodactyly and arch defects of the hands and feet.

Occasionally they may have deafness and ear malformations. Normal intelligence.

Paraclinical examinations

The diagnosis is eminently clinical.

Skin Biopsy. To check for ectodermal dysplasia, it may be useful in some cases.

Syndromogenesis and etiology

The etiology is genetic due to a monogenic mutation with autosomal dominant inheritance, reduced penetrance and variable expressivity.

Literature

Bixler. D., et al.: "The ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome" Clin. Genet., 3:43-45, 1972.

EMBRYOPATHY SYNDROME BY THALIDOMIDE

Synonymy

S. de Wiedemann I.

Syndromography

Clinic

It occurs in newborns whose mothers took thalidomide (alpha-N-phthalidomido glutaramide) during pregnancy, the most critical period being between 37 and 50 days after the last menstruation.

This syndrome was very common between the 50s and 60s when the product was introduced on the market as a commonly used analgesic, which gave rise to a true epidemic, as countless cases occurred. At present it is rare due to the control measures that have been taken.

It affects both sexes equally and is characterized by deformities in the development of the limbs with a variable range between amelia and deformities of the thumbs. Deformities are more frequent in the upper limbs, they are generally bilateral, but usually asymmetric.

There are gradations of defects by reduction of members: hypoplasia or aplasia of the thumbs, polyphalangies and polydactylies or syndactylies.

Auricular defects are also described, including anotia or absence of the external auditory canal, and there may be mild dysmorphisms such as a mesofrontal nevus flamneus or a mustache-like upper lip.

Malformations include: heart disease, stenosis or atresia of the duodenum and/or anus, appendiceal aplasia, absence of the lower lobe of the right lung, renal aplasias and malformations, asplenia and very rarely mental retardation.
Associated are: cranial malformations, hydrocephalus, meningo-myelocele, macrophthalmia or anophthalmia, saddle nose, cleft palate, facial hemangioma.

It is usual for intelligence to remain normal.

Paraclinical examinations

The diagnosis is clinical.

Syndromogenesis and etiology

This syndrome occurred between 1958 and 1962 in Europe and Australia due to the use of thalidomide as an antiemetic drug in pregnant women. In this period, more than 5,000 cases occurred in Germany. Thalidomide causes developmental disorders

embryogenesis of limbs and organogenesis whose severity correlates with gestational age and the doses used.

Literature

Lenz, W. and K. Knapp: "Die thalidomid embryopathie." Dtsch. Med. Wschr., 85:1232. 1962.

Lenz, W.: "Das thalidomid-syndrome." Fortschr. Med. 81:148, 1963.

Trueta, J.: "Care of thalidomide babies: Experience in Germany." Lancet, 2:1162, 1962.

SPORADIC ERYTHROCYTOSIS SYNDROME IN CHILDREN

Synonymy

S. of primary erythrocytosis.

Syndromography

Clínic

In this syndrome, which is characterized by an isolated increase in the mass of red cells, without any known anomaly as a cause of secondary erythrocytosis lacking familial transmission, two groups have been recognized:

1. Cases with pure erythrocytosis without associated growth abnormalities. Normal leukocytes and platelets, occasionally mild splenomegaly.
2. Cases with pure erythrocytosis and complex anomalies of growth and endocrine development. This group may be identical to cases with dysencephalic lesion and erythrocytosis.

Syndromogenesis and etiology

The mechanism and etiology of this syndrome have not been determined.

Literature

Darvey, M.G.; J.R. Lawrence; H. Langer et al. : "Familial erythrocytosis, a report of two cases and a review." Minutes, Haemat. 39:65-74, 1968.

Nathan, M.: "Erythrémies protopathiques et diancéphale." Persse.Med., 39:403-404, 1931.

SCLEROSTEOSIS SYNDROME

Synonymy

S. of cortical hyperostosis-syndactyly.

Syndromography

Clínic

It begins in the infant due to the development of the forehead, although family members usually consult for neurological symptoms. Facies: high forehead, hypertelorism, wide, flat nasal root; mandible square, prognaic; increased cranial circumference, short mesofacies. Syndactyly: symmetrical, between the second and third fingers, at least along the length of the first phalanx; deformation of the index finger with onychodysplasia, radial deviation of the second and third fingers. Nerve compression: Cranial with hearing loss that sometimes reaches deafness and dumbness preceded by recurrent otitis and stenosis of the external auditory canal; ambioply and occasionally blindness due to optic atrophy, papillary edema, convergent strabismus, nystagmus and exophthalmia, facial paresis. Normal intelligence.

Paraclinical examinations

Radiology. General densification of the skeleton, particularly the skull and base of the skull, mandible, ribs, clavicles, reduced maxillary sinuses, long bones, metacarpals, phalanges without diaphyseal narrowing.

Syndromogenesis and etiology

Densification of the compact and spongy region of the bones.

It is transmitted in an autosomal recessive manner.

The etiology is genetic.

Literature

Falconer, A.W. and B.J. Ryrie: *Report of a familial type of generalized osteo-sclerosis with report on pathological changes". Med. Press., 195:12, 1937.

Hansen, H.G. in Opitz-Schmid: Handbuch, der Kindemeikunde, Ed. Springer, Berlin; t.VI, p. 351.

Truswell, A.S.: "Osteopetrosis with syndactyly: a morphologic variant of Albers-Schonberg disease." J. Bone Joint Surg., 40 B:208. 1958.

SHORT STAGE-HIGH IGA-ASTHMA SYNDROME

Synonymy

Asthma syndrome-short stature-high IgA.

Syndromography

Clinic

This syndrome appears in both sexes. It begins between 4-5 months of age, with recurrent asthma attacks. Between attacks the patients are free of respiratory symptoms. At the age of 4-5 years it begins with seasonal episodes of nasal congestion, coryza and nasal itching.

Physical examination. Short stature.

Paraclinical examinations

Blood. Great elevation of IgA globulin; depression of IgM concentration.

Syndromogenesis and etiology

It is related to a genetic defect not yet established.

The etiology is unknown.

Literature

Huntley, C.C.; H.W. Johnson and A.D. Lyerly: “Asthma, short stature and elevated gamma iga globulins.” Amer. J. Dis. Child., 109:358, 1965.

Sly, R.M. and E.M. Heimlich: Identical twins with short stature elevated IgA and asthma." Ann. Allerg., 25:578-86, 1967.

PULMONARY STENOSIS SYNDROME AS A COMPLICATION OF MATERNAL RUBELLA

Syndromography

Clinic

Newborn of a mother who suffered rubella during the gestational period. Has a maximum systolic murmur at the center

pulmonary frequently with extensive irradiation, the ejection sound is not frequently present and the second sound is normal. Other findings appear in relation to congenital rubella syndrome.

Paraclinical examinations

Chest radiology. The cardiac area is normal, there is generally no abnormal prominence of the pulmonary artery and the pulmonary vascular network is normal. When there is a shunt from left to right, there may be cardiomegaly and pulmonary plethora.

Cardiac catheterization. It indicates pressure gradients across areas of obstruction that are not always easily recognizable.

Angiocardiography.

Electrocardiograph. Signs of hypertrophy of the right atrium and ventricle.

Analog studies. In congenital rubella, the virus can be isolated in the throat, blood, urine, cerebrospinal fluid, lens, and other organs that are affected by the disease.

Pathology. Association of pulmonary stenosis with patent ductus arteriosus.

Syndromogenesis and etiology

Rubella virus infection in the mother during the gestational period.

PULMONARY STENOSIS SYNDROME - PERSISTENCE OF THE FORAMEN OVAL

Syndromography

Clinic

Cyanosis, especially on exertion, can appear shortly after birth, but late onset is more common, after puberty or in adult life.

Occasionally the patient bends over. He has inadequate weight gain, repeated lung infections, fatigue and exceptionally dyspnea.

Physical examination. Clubbing of fingers, high-pitched ejection murmur (similar to that seen in pure pulmonary stenosis), single second sound. Administration of amylnitrate decreases the murmur.

Paraclinical examinations

Chest radiology.

Angiography.

Electrocardiograph.

Oximetry.

Syndromogenesis and etiology

It is a congenital heart malformation.

The etiology is unknown.

Literature

Selzer, A.; W.H. Meats; AC Noble et al.: "The syndrome of pulmonary stenosis with patent foramen ovale." Amer. J. Med., 6:3-23, 1949.

PURE PULMONARY STENOSIS SYNDROME

Synonymy

S. of pulmonary stenosis with intact interventricular septum. S. of isolated pulmonary stenosis.

Syndromography

Clinic

The age of incidence ranges between childhood and adulthood. Generally this syndrome is asymptomatic for long periods, but sooner or later fatigue, dyspnea on exertion appear, occasionally chest pain or syncope on exertion, there may be a stabbing sensation.

Physical examination. The normal-looking patient. If the stenosis is very noticeable, it may present with a full moon face and peripheral cyanosis.

Auscultation: strong systolic pulmonary ejection murmur accompanied by auscultable thrill with greater intensity in the pulmonary area and frequently preceded by an ejection pulmonary tone. There may be a splitting of the second lung tone.

Paraclinical examinations

Electrocardiogram. It reveals right ventricular hypertrophy frequently accompanied by high and pointed P.

Chest radiology. Poststenotic dilation of the pulmonary artery with hypertrophy of the right ventricle and atrium is observed. Pulmonary vascularization is decreased.

Cardiac catheterization. Increased pressure gradient at the level of the pulmonary valve. The pressure in the pulmonary artery is normal or low. Systolic ventricular pressure is high. In severe cases the right atrial pressure shows a prominent "a" wave.

Selective angiocardiography. Dilated pulmonary artery is observed, sometimes the abnormal pulmonary valve can be seen.

Syndromogenesis and etiology

It is a congenital heart anomaly. A familial association with an autosomal recessive pattern has been seen in some cases. This syndrome can be stimulated by external compression of the pulmonary artery.

The etiology is unknown.

Literature

Currens, J.H.: TD. Kinney and P.D. White: "Pulmonary stenosis with intact interventricular septum." Amer. Heart J., 30:491-510, 1945.

BLADDER EXTROPHY SYNDROME

Synonymy

Bladder exstrophy sequence.

Syndromography

Clínic

It is a congenital malformation in which the bottom of the bladder appears exteriorized as a mucosal surface with the ureteral orifices excreting urine continuously. It can be accompanied by umbilical and/or inguinal hernias, rectal prolapse, and separation of the pubic bones. Other malformations: imperforation of the anus, cryptorchidism, duplication of the upper urinary tract.

Paraclinical examinations

Clinical diagnosis

Syndromogenesis and etiology

It is caused by a primary defect in the infraumbilical mesoderm that results in a sequence of exposure of the posterior wall of the bladder, incomplete fusion of the genital tubercle, and separation of the pubic rami. The causes that determine the primary defect are not known.

Literature

Smith, D.W.: Recognizable patterns of human malformation. 2nd. ed., Saunders, Philadelphia, 1976, p. 378.

Uson, A.C.; J.K. Lattimer and M.M. Melicow: "Types of ecstrophy of primary bladder and concomitant malformations" Pediatrics, 23: 927, 1959.

Williams, I.: Pediatric Urology. London, Butterworth, 1968. p. 229.

Wilson, J.G.: The Intern. Med. Congr. Ltd.: First Intern. Am. Conference on Cong. Defects. Ed. Lippincott, Philadelphia, 1963, p.113.

FAMILIAR AUTOSOMAL SYNDROME

ADDISON ASSOCIATE

A HYPOPARATHYROIDISM

Syndromography

Clinic

It affects both sexes and begins after the first year of life.

Moniliasis infection (see hypoparathyroidism syndrome) and symptoms and signs of hypoadrenalismno (see Addison syndrome).

It can be associated with keratoconjunctivitis, cirrhosis, Hashimoto' s disease and pernicious anemia.

Paraclinical examinations

Blood. Hypocalcemia, hyperphosphoremia.

Urine. Absence of kidney failure. Decreased excretion of 17-ketosteroids and 11-oxysteroids after ACTH stimulation (see Addison syndrome). Therapeutic response to parathyroidism with acute phosphoric diuresis.

Radiology. Absence of rickets or osteomalacia.

Syndromogenesis and etiology

A possible autoimmune disorder and autosomal recessive inheritance are invoked.

Literature

Supthin, A.; F. Albright and D.J. McCune: “Five cases (three in sibings) of idiopathic hypoparathyroidism associated with moniliasis.” J. Cin. Endocrin. , 3:625-34, 1943.

Taiz, L.S.; C. Zarate and E. Schwartz: "Congenital absence of parathyroid and thymus glands in an infant." Pediatrics, 38:412- -18, 1966.

FARBER SYNDROME

Synonymy

S. of disseminated lymphogranulomatosis.

Syndromography

Clinic

It begins in the first months of life: laryngeal stridor occurs, increased volume of the joints of the the members. Infiltrative skin lesions. Hepatomegaly and physical and mental retardation.

Paraclinical examinations

Blood. There is moderate leukocytosis (15,000-20,000).

Anemia. Cholesterol and total lipids are normal.

Radiology. Osteoporosis. Destructive changes in joints and periarticular calcifications.

Liver biopsy. Normal.

Syndromogenesis and etiology

The etiology is genetic. The basic defect consists of a deficiency of acid ceramidase. It presents autosomal recessive inheritance.

Literature

Farber, S.: "A lipid metabolic disorder-disseminated lipogranulomatosis. A syndrome with similitarity to and important differences from Niemann-Pick and Hand-Schuller-Christian disease". Amer. J. Dis. Child., 84:499-500, 1952. (abstr.)

Sugita, M.; J.T. Dulaney and H.W. Moser: "Ceramidase deficiency in Farber's disease (lipogranulomatosis)". Science, 178:1100- -1102, 1972.

FETAL ALCOHOL SYNDROME

Syndromography

Clinic

The clinical characteristics are variable from growth deficiencies of prenatal or postnatal onset to intelligence defects with I.Q. average of 63. They present fine motor dysfunction, poor coordination and tremor. Irritability in childhood, hyperactivity in schoolchildren. They have Craniofacial dysmorphia with moderate microcephaly, short palpebral fissures, maxillary hypoplasia, short nose, smooth philtrum with thin upper lip. Small distal phalanges with hypoplasia of the nail of the fifth finger. Joint anomalies. Cardiovascular defects with ventricular and atrial septal defects.

Syndromogenesis and etiology

The etiology is prenatal due to ethanol or its bioproducts. At least 4-6 drinks per day are required for the described manifestations to become evident. Most children with all the clinical features of the syndrome are children of alcoholic women who drink more than 8-10 drinks per day. It has been recognized that 2 drinks per day reduces size at birth and sporadic drunkenness in the first trimester of pregnancy has even been related to children who present learning disorders and some of the dysmorphic signs described.

The risk for alcoholic mothers of having children with the syndrome has been estimated at 30 to 50%.

Malformation defects include neuron heterotopia and frank brain malformations.

Literature

Jones, K.L.: "Fetal alcohol syndrome." Pediatrician Review, 8:122- -126, 1986.

FETAL RETINOIC ACID SYNDROME

Synonymy

S. of embryopathy due to retinoic acid.

Syndromography

Clinic

The patient is characterized by presenting a dysmorphic pattern with large defects of the auricular pavilions (microtia or anotia with stenosis of the auditory canal); inclined narrowing of the forehead, micrognathia, depression of the nasal bridge and ocular hypertelorism, U-shaped palate with clefts. It is accompanied by severe cardiovascular malformations (transposition of the great vessels, tetralogy of Fallot, common truncus arteriosus, ventricular septal defects), retroesophageal subclavian artery, hypoplasia of the aortic arch. Malformations of the central nervous system (microcephaly, hydrocephalus, neuronal migration defects and important malformations of structures of the posterior fossa such as cerebellar hypoplasia, agenesis of the vermis, megacisterna). They also have thymus abnormalities.

Paraclinical examinations

Radiology. Accessible parietal sutures are observed in the skull.

sorias

Echocardiography. Diagnosis of citric malformations

tada.

Brain ultrasound, and computed axial tomography and magnetic resonance imaging. Diagnosis of defects of the central nervous system.

Syndromogenesis and etiology

This syndrome is produced by the teratogenic action on the embryo of drugs containing isoretinoic acid or retinoic acid and although there is no evidence of its effect due to use prior to pregnancy, this is likely due to its storage in body fat. Its use in the first trimester of pregnancy produces 22% of spontaneous abortions, 3% of malformed stillborns, and 11% of live births with at least one major malformation.

Literature

Lammer, E.J. et al.: "Retinoic acid embryopathy." N. Engl. J. Med., 313:837, 1985.

Rosa, F.W.: "Teratogenicity of isotretinoin." Lancet, 2:513-515, 1983.

FETAL AMINOPTERIN SYNDROME

Synonymy

S. of embryopathy due to folic acid antagonists.

Syndromography

Clínic

The clinical picture is so characteristic that when it has been seen once it can be easily recognized in the infant.
There are frontal, parietal or both ossification defects, brachycephaly, as well as a dysmorphism characterized by irregular and abnormal hair implantation. Hypoplasia of the supraorbital ridges, the eyebrows are hyperpigmented in their middle part, the patients present significant ocular hypertelorism, exophthalmos, blepharophimosis, very prominent nose, micrognathia, malformed ears with posterior rotation, high and sometimes cleft palate. There is shortening of the limbs especially mesomelic and variable defects of the hands and feet: brachydactyly, syndactyly and hypoplasia of the fingers. Intelligence is normal.

Paraclinical examinations

Radiology. Demonstrate the defects mentioned. In the lateral views of the skull, synostosis of the lambdoid and coronal sutures, very large fontanelles, and hypoplasia of the calvarial bones are also observed.

Syndromogenesis and etiology

The etiology is based on the teratogenic action of drugs such as aminopterin and methotrexate, whose effect as folic acid antagonists produces a significant deficiency of folic acid in the embryo. In high doses they suppress fetal hematopoiesis and can cause liver necrosis and fetal death. Doses of 6-12 mg produce embryopathy.

Literature

Warkany, J.: "Aminopterin and methotrexate: folic acid deficiency." Teratology, 17:353, 1978.

FETAL SYNDROME BY CYTOMEGALOVIRUS (CMV)

Synonymy

Cytomegalovirus S. in the fetus.

Syndromography

Clinic

The symptoms of this syndrome can vary in intensity from the absence of clinical signs to the presence of serious infection. The syndrome is characterized by hepatosplenomegaly, petechiae, jaundice, anemia, 30% of patients are premature. Hydrocephalus or microcephaly. Mental retardation, hyperactivity, seizures, spasticity, hearing loss. Chorioretinitis or atrophy of the optic nerve in 10% of cases.

Paraclinical examinations

Blood. Thrombopenia (60%), hemolytic anemia (60%), elevated serum Igm in (85%).

Cerebrospinal fluid. Proteins in 50% of

cases.

Radiology. Skull (brain calcifications), long bones (metaphysitis).

Liver biopsy. Giant cell hepatitis, portal fibrosis, cholangiitis.

Detection of the virus in urine. Children infected with CMV shed it in the urine for several years although inclusion cells are seen in one third of cases.

Determination of specific antibodies.

Syndromogenesis and etiology

It is produced by the teratogenic effect of cytomegalovirus in the uterus via the transplacental route, in the third or fourth month.

The virus produces vascular necrosis.

Literature

Pass, R.E. et al.: "Out come of symptomatic congenital CMV infection: results of long-term follow-up". Pediatrics, 66:758-762, 1980.

FETAL HDANTOIN SYNDROME

Synonymy

S. fetal by Dilantín.

Syndromography

Clinic

This syndrome is characterized by a variable spectrum of manifestations, the most severe end of the spectrum presents growth abnormalities of prenatal onset that can become accentuated in later months. It is accompanied by mental deficiency and craniofacial dysmorphism with wide anterior fontanelle, metopic ridge, ocular hypertelorism, wide and depressed nasal bridge, short nose, bulging upper lip, and prominent gums. Characteristic of the syndrome is nail hypoplasia, especially in post-axial digits, and hypoplasia of the finger ridges with a pattern of arches on the fingers. In addition, digitization of the thumb, hip dislocation, short neck, teletelia, umbilical and inguinal hernias, pilonidal sinus, hirsutism and malformations such as cleft lip and palate, coloboma, strabismus, congenital heart disease, pyloric stenosis, duodenal atresia, renal malformations, genital malformations, holoprosencephaly sequence can also be found.

Paraclinical examinations

Radiology. Hypoplasia of distal phalanges.

Dermatoglyphs. Digital arches and hypoplasia of the ridges.

Brain and abdominal ultrasound. Look for malformations

mations described.

Syndromogenesis and etiology

It is due to the teratogenic effect of hydantoin.

The risk of the syndrome in fetuses exposed to idantoin is 10% and the risk of having any of the signs and symptoms is an additional 33%. Numerous studies suggest that fetal susceptibility to the teratogen hydantoin depends on the fetal genome and that an inherited detoxification defect may contribute.

Literature

Monson, R.R. et al.: “Diphenylhydantoin and selected congenital malformations.” N. Engl. J. Med., 289:1049-1052. 1973.

Strickler, S.M. et al.: "Genetic predisposition to phenytoin-induced birth defects." Lancet 2:746-748, 1985.

FETAL RUBELLA SYNDROME

Syndromography

Clinic

Cataracts, deafness and persistent ductus arteriosus are the fundamental clinical manifestations. Growth deficiency of prenatal onset, microcephaly, other ocular defects such as glaucoma, corneal opacity, chorioretinitis, microphthalmia, strabismus; and cardiovascular defects such as peripheral pulmonary, septal, and vascular stenosis due to an increase in the intima and media layers of large arteries. Hepatos- plenomegaly, obstructive jaundice, metaphyseal osteolysis. Mental retardation of various degrees in correspondence with the trimester of gestation of the infection.

Paraclinical examinations

During the neonatal period:

Hematological study. Hemolytic or hypoplastic anemia, thrombocytopenia.

Cerebrospinal fluid. Increase in proteins and ce-

lulas.

Determination of serum IgM. 20% of fetuses have increased cord blood (more than 20 mg/dL). 50%, months after birth, have figures 4 times higher than that of the mother. Isolation of the virus in pharyngeal swab, cerebrospinal fluid, blood, urine, bone marrow, liver, in ear secretions (otitis). Determination of specific antirubella IgM. such as Echocardiography, ultrasound and PEATC. After one year of age, for the diagnosis of the indicated malformations.

Syndromogenesis and etiology

The teratogenic effect of the rubella virus is produced by inhibition of mitosis, inflammation and loss of critical cells during organogenesis, as well as chronic inflammation in various organs.

If the infection occurs in the first month, 50% of the embryos are affected, and 5-10% when it occurs at 20 weeks.

When cataracts are present, the infection must have occurred between 26 and 57 days of pregnancy. The other defects occur between days 126 and 131.

Literature

Hardy, J.B "Clinical and developmental aspects of congenital rubella." Arch Otolaryngol., 98:230-236, 1973.

Meda, K. et al.: "Congenital rubella syndrome: correlation of gestational age at time of maternal rubella with type of defect."

J. Pediatr., 94:763-765, 1979.

FETAL SYNDROME DUE TO TOXOPLASMOSIS

Synonymy

S. of toxoplasmosis in the fetus.

Syndromography

Clinic

The spectrum of findings is wide from stillborn newborns to apparently healthy but infected children. Prematurity is common. In the syndrome, 80% have mental retardation and seizures. Hydrocephalus or microcephaly, unilateral or bilateral chorioretinitis with microphthalmia, secondary glaucoma. Hepatosplenomegaly, jaundice, purpura, maculopapular skin rash. Pneumonitis and carditis several weeks after birth. Children may be born apparently healthy and later present with seizures, chorioretinitis, mental retardation or deafness.

Paraclinical examinations

Detection of antitoxoplasma antibodies. If the child is not infected and the antibodies have been acquired passively through the placenta, the titers will decrease by 50% each month. If there really was an embryofetal infection, the titers in the child increase in serial determinations.

Determinations of IgM and antitoxoplasma IgM by fluorescent antibodies. Isolation of the parasite in cerebrospinal fluid where there is also mononuclear pleocytosis and high protein levels.

Skull radiology. Brain calcifications in periventricular regions and basal ganglia.

Syndromogenesis and etiology

The cause is the teratogenic effect of the protozoan Toxoplasma gondiu on the fetus from a mother who is infected for the first time.

once during pregnancy. The proliferation of the microorganism causes granulomatous inflammation and necrosis of fetal tissues, especially the brain. Congenital toxoplasmosis does not recur in subsequent pregnancies.

Literature

Desmontes, G. and J. Couvreur: "Congenital toxoplasmosis: a prospective study of 378 pregnancies." N. Engl. J. Med., 290:1110-1116, 1974.

TRIDIONA FETAL SYNDROME

Synonymy

S. fetal by trimethadione.

Syndromography

Clinic

It is characterized by multiple defects with a wide range of variation at the beginning due to low stature and hypodevelopment of prenatal onset, mental defect with dyslalias, pattern of dysmorphic craniofacial signs: moderate brachycephaly, discrete hypoplasia of the middle facial third, short nose with anteverted nostrils, prominent forehead, V-shaped eyebrows, epicanthus, eyelid ptosis, strabismus; high arched palate, micrognathia, overcoiling of the helix and other ear defects; congenital malformations from cleft lip and palate to septal defects, tetralogy of Fallot, ambiguous genitalia, postpadias, clitoral hypertrophy. Other congenital defects such as facial hemangioma, membranous neck, transposition of the great vessels, hypoplastic heart, pyloric stenosis, kidney anomalies, inguinal and umbilical hernias, hip dislocation, and visual deficit and hearing loss may also be observed.

Paraclinical examinations

Necessary to detect the malformations described.

Echocardiography, abdominal ultrasound and others.

Syndromogenesis and etiology

The use of trimetadione and paramethadione during pregnancy has been related to the teratogenic effect of this drug and can cause spontaneous abortions in 13% and newborns with the syndrome or one of its major malformations in 83%.

The frequency and severity of the syndrome associated with the use of tridione are sufficient elements to consider elective abortion in pregnant women who have consumed the drug, especially during the first trimester.

Literature

Feldman, G.L.; D.D. Weaver and E.W. Lovkien: The fetal trimethadione syndrome". Am. J. Dis. Child., 131:1339-1392, 1977.

Gorman. J.: A. Lowal and KH. Ehlers: Trimethadione and humans teratogenesis." Teratology, 3:349-352, 1970.

WARFARIN FETAL SYNDROME

Synonymy

S. of warfarin embryopathy.

Syndromography

Clinic

The syndrome presents a special dysmorphic pattern with nasal hypoplasia and nasal bridge, the nasal tip is pinched;
nail hypoplasia and shortening of the fingers.

Low birth weight and marked psychomotor retardation.
Various malformations of the central nervous system may occur, including: microcephaly, hydrocephalus, agenesis of the corpus callosum, Dandy-Walker malformation, optic atrophy, microphthalmia, among others, as well as congenital heart diseases. It presents a skeletal defect in 100% of the outlined cases characterized by non-calcified epiphyses and instead a granular stippling particularly in the lumbosacral region and iliac wings, head of the femur and in the calcaneus.

Paraclinical examinations

Radiology. In the newborn and before one year of age, since the stippling disappears after this age and they must be selective of the lumbosacral spine, AP and L pelvis as well as L of the foot, to observe stippling of the calcaneus.

Brain ultrasound. For diagnosis of central nervous system anomalies.

Syndromogenesis and etiology

This syndrome is related to the teratogenic effect of coumarin derivatives that act as anticoagulants.
A direct teratogenic effect on the central nervous system related to the inhibition of calcium-binding proteins during a critical embryonic period of ossification has been suggested for the rest of the clinical picture and especially for ossification defects. This syndrome is a phenocopy of chondrodysplasia punctata that has genetic etiology.

The newborns of 5% of pregnant women who consume this drug present the complete syndrome, 30% have mental retardation, 15% have eye defects and 50% have dystrophy of the fingernails and toenails.

Literature

Hall, J.G.; R.M. Pauli and K.M. Wilson: "Maternal and fetal sequalae of anticoagulation during pregnancy." Am. J. Med., 68:122- -140. 1980.

Warkany, J.: "Warfarin embryopathy." Teratology, 14:205-209, 1976.

FOLLING SYNDROME

Synonymy

S. of phenylpyruvic oligophrenia. S. of phenylpyruvic imbecility. S. of phenylpyruvic idiocy. S. of phenylketonuria.

Syndromography

Clínic

This syndrome was first described in 1934 by Folling.

The child is born normal and within a few days intense vomiting appears, which is sometimes mistaken for pylorostenosis, and the urine has a strange mouse-like smell. Irritability, seizures.

Intelligence: around 3 months the IQ begins to decline, and at 3 years it is around 40%.

Physical examination. Skin: rough, later eczematous (20%);
The hair is depigmented and lightened (62%) and 20% are dark-skinned. Tendency towards photosensitivity that causes photodermatosis; hyperhidrosis; Secondary eczematization of the skin, which predisposes to intertrigo and follicular keratosis. Central nervous system: minimal microcephaly (2 cm smaller than normal); Between 6-18 months, petit mal attacks begin, which cease spontaneously before reaching adulthood. They present hypertonia (70%) or hypotonia (20%); Tendon hyperreflexia (66%) sometimes reaching clonus, agitation, tremors. The child becomes fearful with tantrums and destructive behavior (32-90%); occasionally, due to spastic cerebral lysis and sometimes dental enamel hypoplasia.

Paraclinical examinations

Blood. In the normal infant, phenylalanine is 2-4 mg/100 mL. In this syndrome it exceeds 20 mg/100 mL.
Hypophosphatemia, decreased alkaline reserve, hypoglycemia on an empty stomach.

Urine. Various metabolites of phenylpyruvic acid are excreted and react with iron chloride.

Electroencephalogram. With spikes-waves, dysrhythmia and petit mal variant.

Pneumoencephalogram. Indicates brain atrophy.

Syndromogenesis and etiology

The deficiency of phenylalanine-hydroxylase prevents the transformation of phenylalanine into tyrosine and accumulates in tissues, producing hair depigmentation, eczema and lesions of the central nervous system, by altering the cellular environment, inhibiting other enzymes, which produces irreversible lesions.
It is an innate disorder of metabolism in the liver.

It is transmitted in an autosomal recessive manner; Heterozygotes, although they have increased phenylalanine in their blood, do not have brain damage. The frequency in the white race is one case per 20,000 births.

The etiology is genetic.

Literature

Folling, A.: "Ueber Auscheidung von Phenylbrenztraubensaure in den Ham als Stoffwechselano-malie in Verbindung mit imbezilitaet." Physiol. Chem., 227:169, 1934.
Jervis, G.A.: "The genetics of phenolpyruvic oligophrenia." J. Ment. Sci. 85:719, 1939.
Knox, W.E.: In Stanbury-Wyngaarden-Frederickson: Metabolic basis of inherited disease. 3 a.m. ed., Ed. McGraw-Hill. New York, 1972. p. 266.

FORBES SYNDROME

Synonymy

S. of borderline dextrinosis and debranched glycogen. S. enzymatic limit dextrinosis. Cori type III glycogenosis syndrome.
S. of amylo-1,6-glucosidase deficiency.

Syndromography

Clínica

It affects both sexes and debuts early in childhood.

Patients have a craving for sweets and carbohydrates, without which they present hypoglycemic attacks with occasional seizures.
Moderate muscle weakness, easy fatigue.
Harris syndrome (see this one). Moderate mental retardation, growth delay that normalizes at puberty. Normal intelligence.

Physical examination. Doll face. Marked hepatomegaly that disappears as the child grows (after approximately four years).

Paraclinical examinations

Blood. Amylo-1,6-glucosidase deficiency in leukocytes and erythrocytes. Hypoglycemia, acetonemia, hyperlipidemia.

Hypercholesterolemia. Little hyperglycemic response to adrenaline and glucagon. Increased glycogen in red blood cells, liver and muscles.

Liver and muscle biopsy. Increased glycogen, steatosis, absence of amylo-1,6-glucosidase activity.

Electrocardiogram and electromyogram. Normal.

Syndromogenesis and etiology

This syndrome is caused by the accumulation of glycogen in the liver, heart and muscles. Stored glycogen is structurally abnormal because of its short chain (called limit dextrin, hence the name dextrinosis). This is due to the lack of amylo-1,6-glucosidase (debranching enzyme) in the muscle and liver, which causes an alteration between the side chain and the main chain. The persistence of this biochemical defect noticeably inhibits the phosphorylation of the 4 glucose unit and a short-chain dextrin "limiting" residue is produced as a residue.

It is transmitted by autosomal recessive inheritance.

Literature

Forbes, G.B.: "Glycogen storage disease; report of a case with abnormal glycogen structure liver and skeletal muscle." J. Pediat., 42:645-53, 1953.
Howell, R. in Stanbury-Wyngaarden-Frederickson: The metabolic basis of inherited disease. McGraw-Hill, 3 ed., 1972, p. 95.
Illingworth, B.; T. Cori and C.F. Cori: “Amylo-1, 6-glucosidase in muscle in generalized glycogen storage disease." J. Biol. Chem., 218:123, 1956.
Pearson, C.M.: "Glycogen metabolism and storage diseases of type II, IV and V." Amer. J. Clin. Path. 50:29-43. 1968.
Williams, C. and J.B. Field: "Studies in glycogen storage disease: II. Limit dextrinosis to a genetic study". J. Pediatr., 72:214, 1968.

FRAGILE X SYNDROME

Synonymy

S. of mental retardation linked to the X chromosome and macrorchidism. S. X chromosome marker. S. by Martin Bell. S. of Fraxa.

Syndromography

Clínica

The clinical manifestations of this syndrome begin in childhood due to a psychomotor delay, mainly in the development of language and certain manifestations of behavior and social relationships similar to those observed in children with autism. Language abnormalities include semantic errors, mental processing defects, anxiety, perseveration, echolalia.

At birth, weight, height, and head circumference are normal or greater than the 75th percentile. They have a characteristic facial dysmorphism: the face is elongated, the chin is large, the ears are prominent and show alterations typical of connective tissue defects; high palate, hypotonia, hypermobility

ligamentous, flat feet, scoliosis, mitral valve prolapse, dilation of the ascending aorta, thin skin. 60% of patients present epilepsy and post-pubertal males have macrorchidism, which together with facial dysmorphism constitute physical signs of great value in clinical diagnosis. The phenotypic variations observed, including the gradations of the severity of mental retardation, have suggested that every man with non-specific perinatal mental retardation, psychosis or without an apparent cause of this defect should be checked whether or not it is this syndrome.

Paraclinical examinations

Karyotype. Culture of leukocytes in folate-poor media induces the expression of the fragile site in Xq27.3. The analysis of 100 metaphases should express between 2-30% more of these gaps.

Molecular studies. Southern blot and/or PCR analysis with the appropriate probe allow the presence of the FMR 1 mutation to be identified, both in the sick male and in the heterozygous females.

Syndromogenesis and etiology

The etiology is genetic, due to a dynamic mutation that affects the FMR 1 gene in the number of repeats (CGG)n at its 5 end.

Normal people have a range of repetitions (CGG)n between 6 and 53, but from 43 repetitions it can increase its number to 200 and become a premutation that, although it does not produce phenotypic effects, people who have it can transmit it to their offspring: if it is a man, to 100% of its daughters who will be carriers, if it is a woman, the number of repetitions (CGG)n expands to more than 200, giving rise to complete mutation that can affect male children and even their carrier daughters.

The Xq27.3 fragilities observed in the metaphases of sick patients correlate with the severity of the clinical manifestations.

The complete mutation prevents transcription of the gene (the armm is absent), however, the function of the protein expressed by the normal gene has not been clarified and therefore the effect of its absence to explain the neurological manifestations of the affected male has not been clarified.

Bibliography

Lantigua, A: Fragile X syndrome. Conference. Updated course on Clinical Genetics Higher Institute of Medical Sciences of Havana, 1993.

FREEMAN-SHELDON SYNDROME

Synonymy

S. of the whistler facies. S. of cranio-carpotarsal dysplasia. Buccal-facial-digital S.

Syndromography

Clinic

It mainly affects the face, hands and feet. The facies is characteristic of a mask and with a small mouth that gives the appearance of a

Abnormality in the development of the sublingual frenulum.

Pseudoclefts in the upper lip, tongue and bony palate.

High palate, small tongue, limited palate movements with nasal voice.

Deep eyes, wide nasal bridge, telecanthus, epicanthus, strabismus, blepharophimosis. Small nose, hypoplastic nasal wings with coloboma and long filter.

H-shaped skin folds on the chin. Flexed fingers with thick skin on the flexor surface of the proximal phalanges, ulnar deviation of the hands, club feet, vertical talus, contractural fingers and cortical big toe.

Postnatal growth deficiency, hernias, dislocation of

hip.

Birth almost always occurs in the pelvic region and delivery is difficult. They make little progress in development due to frequent vomiting and dysphagia. Mental retardation is not a common characteristic.

Paraclinical examinations

Radiology. Face: protuberance of the facial bones, very inclined anterior cranial fossa.

Syndromogenesis and etiology

It is a genetic defect due to a mutation of autosomal dominant inheritance, and genetic heterogeneity may exist since families with evidence of autosomal recessive inheritance have been reported. Although the underlying defect is not known, the clinical features appear to be secondary to increased muscle tone.

Literature

Antley, R. M. et al.: "Diagnostic criteria for the whistling face syndrome." Birth Defects., 11:161, 1975.

FRONTONASAL SYNDROME

Synonymy

S. of frontonasal dysplasia. S. of midfacial cleft. S. sequence of frontonasal dysplasia.

Syndromography

Clinic

This syndrome can be expressed with great severity but there are also mild cases with few defects. It is characterized by ocular hypertelorism. Lateral displacement of the internal edges. A widow's peak appears on the forehead at the hairline. This dysmorphic sign may be the only one in mild cases and is due to a hidden bifid skull. The nose can range from a small depression at the tip to a completely divided nose with hypoplasia or absence of the prolabium and premaxilla with a medial cleft lip. The nasal root is wide. Other defects are: accessory nasal appendages, midline dermoid lip0ma, microphthalmia, deafness, mental deficiency. Lipoma of the corpus callosum. Most patients have normal intelligence.

Paraclinical examinations

Skull and face radiology, brain ultrasound or computed tomography. To confirm the indicated defects.

Syndromogenesis and etiology

Of unknown etiology, however the pathogenesis is related to an initial defect in the morphogenesis of the he-

cerebral hemispheres from which the cascade of anomalies or malformation sequence described occurs.

Literature

Sedano, HD. et al.: "Frontonasal dysplasia." J. Pediatr., 76:906, 1970.

FUCCOSIDOSIS SYNDROME

Syndromography

Clinic

It is a disease of postnatal onset, children are normal at birth. The first symptom is the arrest of psychomotor development. At least four forms of fucosidosis are described that are clinically differentiated by the severity of the neurological deterioration and the coarseness of the features:

Type I. It is the most severe form, psychomotor development stops after 10 months, followed by spasticity, tremors, loss of contact with the environment, muscle weakness, hypotonia, decortication rigidity, significant hepatosplenomegaly and recurrent respiratory infections.

Type II. It follows in order of clinical severity; Symptoms begin at 18 months, but in this type there is a characteristic coarse facies (prominent forehead, hypertelorism, very bushy eyebrows, thick lips and tongue). The rest of the clinic is the same as type I.

Type II. It is the least severe, the progress is slower and it presents skin lesions especially in the pubic area (angiokeratomas) and the skin is thick.

Type IV. It is similar to III but dryness of the skin predominates and angiokeratomas do not appear.

Paraclinical examinations

Radiology. Vertebral column: ovoid vertebral bodies with flattening, irregular margins and a peaked projection in lateral view. Coarse bone trabeculae.

Skin biopsy. Deposits of eosinophilic material between the dermis and epidermis.

Electron microscopy. Numerous membranous vacuoles in all tissues.

Urine. Excess oligosaccharides.

Determination of enzymatic activity of a-L-fucosidase. Deficient in all tissues, leukocytes, serum and in fibroblast culture.

Electrolytes in sweat. Chlorine is increased to 2-5 times its normal value.

Syndromogenesis and etiology

Of genetic etiology with autosomal recessive inheritance. This syndrome consists of a defect in the lysosomal enzyme α-L-fucosidase with abnormal accumulation in almost all cells of the body. Clinical heterogeneity could correspond to genetic heterogeneity due to mutations that affect the same enzyme (allelic mutations).

Literature

Willems, P.I. et al.: "Intrafamilial variability in fucosidosis." Clin. Genet., 34:7-14, 1988.

G-SYNDROME

Synonymy

S. of hypertelorism with esophageal anomaly and hypospadias.

Syndromography

Clínica

These patients present a characteristic facial dysmorphism with significant hypertelorism, flattening of the nasal root, prominent parietal ridges, dolichocephaly, large anterior fontanelle, narrow deviated palpebral fissures, epicanthus, inverted nostrils, micrognathia, and dysplastic ears with posterior rotation. Wide or bifid uvula, shortening of the sublingual frenulum.

Genital defects do not exist in females and in males they can vary from mild hypospadias to bifid scrotum with scrotal hypospadias and curvature of the penis, such that the tip of the glans can touch the anterior edge of the anus.

There may be an imperforate anus with a rectourethral fistula.

These patients have difficulty swallowing, causing choking crises, coughing and cyanosis, and gastroesophageal reflux.

The clinical manifestations described together with a hoarse or strident cry make the diagnosis. In chronic cases there is often bronchiectasis.

Paraclinical examinations

Cineradioscopic studies. Neuromuscular dysfunction of the swallowing mechanism with bolus entry up to the middle of the tracheobronchial tree.

Syndromogenesis and etiology

The basic defect is unknown, but the midline organs and structures are mainly affected. It is a syndrome of genetic etiology with autosomal dominant inheritance limited to sex or with inheritance linked to the X chromosome, since in females there is great clinical heterogeneity and no genital defects are observed.

Literature

Van Bieroliet, J.P. and J.D. van Hemmel: “Familial occurrence of the G-syndrome". Clin. Genet., 7:238-244, 1975.

GALACTOSEMIA SYNDROME

Syndromography

Clinic

It affects both sexes; It begins a few days or weeks after ingesting milk. There is vomiting, diarrhea, dehydration, hypoglycemic crisis. Difficulty growing. There are fulminant cases that can lead to death.

In moderate cases: hypotomy, lethargy, severe mental and neurological manifestations. In mild cases only milk intolerance.

Physical examination. Jaundice, hepatomegaly between 4 to 8 weeks of life, cataracts.

Paraclinical examinations

Blood. Increased galactose, hypoglycemia (especially during attacks). Abnormal galactose tolerance test.
Test for determination of galactose-1-phosphate- -uridyl-transferase deficiency in red blood cells.

Urine. Galactose, albumin, aminoaciduria (various amino acids).

Syndromogenesis and etiology

Inability to metabolize galactose due to galactose-1-phosphate-uridyl-transferase deficiency.

It is transmitted by autosomal recessive inheritance. The gene (GALT) is located at 9p13 and there are allelic variants.

Literature

Von Reuss, A.: "Zuckerausscheidung im Sauglingsalter." Vienna. Med. Wschr., 58:799-803, 1908.

Isselbacher, K.J.: Galactosemia. In Stanbury, J.B., J.B. Wyngaarden, and D. S. Frederickson: Metabolic basis of inherited diseases, ed. 2. McGraw-Hill, New York, 1966.

Reichardt, J.K.V and S.L.C. Woo: "Molecular basis of galactosemia: mutions and polymorphism in the gene encoding human galactose-1-phosphate uridytransferase", Proc. Nat. Acad. Sci, 88:2633-2637. 1989.

GARROD SYNDROME

Synonymy

S. of alkaptonuria. S. of ochronosis.

Syndromography

Clinic

It affects both sexes. The manifestations are more severe in men. It begins a few days after birth. There is:

1. Episodes of black urine when standing up or if the urine becomes alkaline (diapers turn black when washed). Black pigment is also excreted with sweat.
2. Arthropathy, back pain, stiffness, which appear in the fourth decade, progressively affecting several joints until total disability in 15 6 20 years. Severe pain in the pubic symphysis. Frequent disc herniation.
3. Renal and prostate lithiasis around the fifth decade.
4. Cardiovascular injury, hoarseness, occasional deafness.

Physical examination. Blue-black pigmentation of the sclera and cartilage of the ear (which also hardens, a sign that appears in the second decade), occasionally of the tip of the nose and tendons. Signs of arthropathy.

Paraclinical examinations

Urine. Determination of homogentisinic acid.

Radiology. Narrowing of the intervertebral spaces, collapse and calcification of the intervertebral discs, degenerative changes in large joints.

Chromatography.

Syndromogenesis and etiology

Deficiency of the enzyme homogentisinic acid oxidase.

It is transmitted by autosomal recessive inheritance.

Literature

Garrod, A.E.: “About alkaptonuria". Lancet, 2:1484-86, 1901.

McKusick, V.A.: Heritable disorders of connective tissue. ed. 3, St. Louis, Mosby, 1966.

GORLIN-CHAUDRY-MOSS SYNDROME

Syndromography

Clinic

This syndrome is characterized by craniosynostosis, hypoplasia of the midface, hypertrichosis, and anomalies of the eyes, teeth, heart, and external genitalia.

Patients have short stature, slight forward bending of the head when walking, hypertrichosis is noticeable in the arms, legs and back, and the hairline is lowered.

They have difficulty opening or closing their eyes completely, they may have coloboma of the upper eyelids, microphthalmia and cone hyperopia, oblique palpebral fissures, nystagmus when looking laterally, limited upward gaze. Astigmatism, corneal bedsores. Sometimes conductive hearing loss. Dental defects are characterized by malocclusion, high and narrow palate, hypodontia, microdontia and abnormalities of tooth shape.

There is patent arteriosus, hypertrichosis, hypoplasia of the labia majora.

Paraclinical examinations

Skull radiology. Synostosis of the coronal suture, brachycephaly, maxillary and nasal bone hypoplasia, ocular hypertelorism, lordosis of the petrous ring, clival hypoplasia and elevation of the lesser wings of the sphenoid.

Eye exam.

Syndromogenesis and etiology

The etiology is unknown although McKusick codes it as a probable autosomal recessive inheritance since it has been described in two brothers. From 1960 to 1962 there were no other cases in the medical literature.

Bibliography

Gorlin, R.J. et al.: "Craniofacial dysostosis, patent ductus arteriosus, hypertrichosis, hypoplasia of labia majora, dental and eye anomalies." J. Pediatr., 56: 778-785, 1960.

Gorlin, R.J. et al.: "Hypoplastic hypocalcified enamel, onycholysis and functional hypohidrosis." Modern Medicine, Nov. 27, 1972; 104.

GORLIN-COHEN SYNDROME

Synonymy

S. of frontometaphysal dysplasia.

Syndromography

Clinic

It is clinically characterized by coarse facies with a broad nasal bridge and prominent supraorbital ridges, incomplete sinus development. Dentition defects: anodontia, delayed eruption and retained deciduous teeth.

High palate, small jaw with decreased angle.

Flexion defect of the fingers, wrists, elbows, knees and hips. Arachnodactyly with long phalanges, thoracic deformity, winged scapulae. Conductive and sensorineural hearing loss that tends to progress. Weakness of muscles in the arms and legs, especially hypothenar and interossei of the hands. Patients may present some degree of mental retardation.

Paraclinical examinations

Radiology. Face: thickening of the orbital rim, absence of frontal sinuses, short maxilla, increased skull base, marked hypoplasia of the angle, condyle of the mandible and prominent antigonial notch.

Skull: enlargement of the foramen magnum.

Vertebral column: abnormalities of the odontoid process of the axis and atlas, flattened lumbar vertebrae, fusion of

second and third vertebrae and subluxation of third and fourth.

Scoliosis.

Long bones: increased density in the metaphysal region that produces a deformity that resembles an Erlenmeyer flask.

Pelvis: flattening of the iliac bones and coxa valga.

Tőrax: ribs and vertebrae with irregular contours.

Hands: carpal bones eroded and fused.

Syndromogenesis and etiology

Of monogenic genetic etiology with recessive X-linked inheritance, it presents severe manifestations in males and female carriers are moderately affected. The basic defect is not known.

Literature

Fitzsimmons, J.S. et al.: "Frontometaphyseal dysplasia. Further delineation of the clinical syndrome." Clin. Genet., 22:195- -197, 1982.

Gorlin, R.J. and B.B. Winter: "Frontometaphyseal dysplasia. Evidence for X-linked inheritance". Am. J. Med. Genet., 5:81-88, 1980.

CHRONIC DELAINEANCE GRANULOCrTOPENIA SYNDROME

Synonymy

S. of chronic benign granulocytopenia of childhood. S. de Vahlquist-Gasser.

Syndromography

Clinic

There is no family history. It is established in childhood or at an early age.

Minor but recurrent infections have been reported: paronychia, gingivitis, ulcerations, furunculosis and respiratory system infections.

During the intervals of the seizures, the child's appearance is normal. Lymph nodes may or may not be palpable or moderate splenomegaly may appear. Children tend to become normal after the age of 4.

Paraclinical examinations

Blood. Moderate leukopenia, absolute and relative neutropenia with values less than 1000, especially neutrophils.

Relative lymphocytosis.

Normal erythrocytes and platelets. The reaction of leukocytes to epinephrine is variable, occasionally it is normal.

Protein electrophoresis. Normal.

Medulogram. Normal cellularity or there is a slight increase. There is variable lymphocytosis. The erythrocyte series is normal and the myeloerythroid index is also normal, except for a total absence of neutrophil maturation. Megakaryocytic series and normal histiocytes.

Syndromogenesis and etiology

There is a non-fatal leukocytogenesis disorder.

miliary.

The etiology is unknown.

Literature

Hotz, A.: "Zur Differentialdiagnoseagranulocytose-Leukamie." Ztschr. Kinderh. 65:529-40, 1949.

Zuelzer, W.w. and M. Bajoghli: "Chronic granulocytopenia in childhood", Blood, 3: 359-74. 1964.

HAJDU-CHENEY SYNDROME

Synonymy

S. of cheney, S. of acrosteolysis. S. of arthro- -dento-osseous isplasia.

Syndromography

Clinic

The clinical manifestations begin postnatally in childhood in relation to changes in the hands and pain, presenting short stature, kyphoscoliosis, distal shortening of the fingers and nails, and early loss of teeth due to alveolar resorption.

Patients are weak and may present pathological fractures. They have thick, straight hair with prominent eyebrows and eyelashes, and low-set ears with prominent lobes. Wide nose and small jaw at the expense of its branches.

Joint laxity.

Paraclinical examinations

Radiology. Skull: cranial bones present suture ossification failures, elongation of the sella turcica, development of basilar impressions and bathrocephaly. Decreased mandibular branches. Biconcave vertebrae, kyphoscoliosis. Hands with acrosteolysis and deviation of the fingers. Shortening of the distal phalanges. Crowding of the carpal bones.

Syndromogenesis and etiology

It is a syndrome of genetic etiology that is transmitted by autosomal dominant inheritance. Sporadic cases appear to represent new mutations.

The basic defect is not known.

Literature

Herrmann, J. et al.: "Arthro-dento-osteo-dysplasia (Hajdu-Cheney syndrome). Review of a genetic acro-osteolysis syndrome." Z. Kinderheilkd. 114:93-97, 1973.

DE HAUWERE SYNDROME

Synonymy

S. of hypertelorism-dysplastic iris-mental retardation.

Syndromography

Clinic

It is noticeable at birth and is characterized by orbital hypertelorism, motor delay and psychological retardation, moderate sensory deafness, dilation of cerebral ventricles, generalized muscular hypotonia and pelvic anomalies.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance, with great expressiveness.

The etiology is genetic.

Literature

Alkemade, P.P.H.: Dysgenesis mesodermalis of the iris and the cornea. Assen the Netherlands. 1969. Ed. Van Gorcum (cited by De Hauwere).

De Hauwere, R.C.; J.G. Leroy; K. Adriaense et al.: "Iris dysplasia, orbital hypertelorism and psychomotor retardation: A dominantly inherited developmental syndrome." J. Pediatr., 82:679, 1973.

Gillespie, FD.: "Aniridia cerebellar ataxia and oligophrenia in siblings." Arch. Opthal., 73:338, 1965.

MILIARY HEMANGIOMA SYNDROME OF THE NEWBORN

Synonymy

Diffuse neonatal hemangiomatosis.

Syndromography

Clínica

It can be seen in both sexes. A large number of hemangiomas scattered over the skin and mucosa are evident at birth; Visceral hemangiomatosis with arteriovenous communications, high-output congestive heart failure, plateletopenia with hemorrhages and involvement of the central nervous system. It is accompanied by dyspnea, tachycardia and jaundice. Most die in early childhood from infection or congestive heart failure secondary to hepatic angiomas with arteriovenous fistulas. Some cases have died due to central neurological or pulmonary complications.

Paraclinical examinations

Biopsy.

Angiography.

Pathology. Hemangiomas are observed in the skin, mucosa and practically in all organs such as liver, spleen, mesentery, pancreas, trachea, central nervous system. There may be some invasion with destruction of normal tissue, but there is no evidence of malignancy.

Syndromogenesis and etiology

There is no evidence of hereditary transmission (insufficient number of cases studied). A possible relationship with Rendu-Osler-Weber syndrome is proposed.

Literature

Burman, D.: P.W.A. Mansel1, and R.P. Warin: "Miliary hemamgiomata in the newborn." Arch. Dis. Child., 42:193-97, 1967. Pediatric Clinics of North America. v. 3. 1983. Pediatric Dermatology I. p. 471-472. Interamerican Publishing House.

HEMIHYPERPLASIA SYNDROME

Syndromography

Clinic

This term identifies the increased growth of one or more parts of the body. It can involve half the body, a limb, one side of the face, or combine some of them.

This asymmetric and disproportionate growth is due to cell proliferation and not to an increase in volume or size. cell loss, hence the most correct term to refer to this alteration is hemihyperplasia and not hemihypertrophy.

To establish the diagnosis of body asymmetry, hemiatrophies must first be excluded.

It is more common on the right side and varies widely in extent and severity. Normal asymmetry exists, although it is not always apparent, and can only be determined by appropriate measurements. Hemihyperplasia syndrome can take two clinical forms:

1. Total: affects the musculoskeletal system and the visceral organs of the affected side.
2. Limited: which in turn presents two varieties, segmental and crossed. The possible associated manifestations are numerous, including skin alterations, malformations of hands and feet, spinal column, genitourinary and neuromuscular. Occasionally they are associated with tumors (Wilms tumor).

Hemihyperplasia may be accompanied by an increase in the volume of the kidneys, testicles, ovaries, and adrenals. The nervous system may also be involved, including hemimegalencephaly.

Paraclinical examinations

The diagnosis is fundamentally clinical by anthropometry, but some tests are useful.

Radiology (bone survey).

Ultrasound and computed axial tomography. The first should be performed during the evolution in the abdomen to investigate nephroblastomas.

Karyotype.

Syndromogenesis and etiology

This syndrome can be:

Isolated: with a frequency of 1 per 86,000. Females are affected twice as often as males.

Associated: it is also observed in the following syndromes:

S. Beckwith-Widemann

S. de Klippel-Trenaunay-Weber.

S. of neurofibromatosis.

S. de Langer-Giedion.

S. of enchondromatosis.

S. de Maffucci.

McCune-Albright S.

S. of Proteus.

It is possibly of multifactorial etiology: nervous, vascular and lymphatic, endocrine and chromosomal abnormalities.

The etiology is heterogeneous.

Literature

Curtius, F.: "Kongenitaler, partieler Riesenwuchs mit endokinenstoerungen'". Dtsch. Arch. Klin. Med., 147:310, 1925.

Fraumeni, J.F. Jr.: "Hemihypertrophy. In Birth Defects Encyclopedia. M. L. Buyse, Cambridge, Blackwell Scientific, 1990, p. 855.

Lenstrup, E.: "Eight cases of hemihypertrophy." Acta Paed., 6:205. 1926.

Meckel, J.E: Ueber die Seitliche Asymmetrie im tierischen Korper. Anatomische physiologische Beobachtungen und Untersuchungen. Halle, Renger, 1822, p. 147.

Ringrose, R.E.; J.T. Jabbour and D.K. Keele: "Hemibypertrophy." Pediatrics, 36:434-48, 1965.

Ward, J. and H.H. Lerner: "A review of the subject of congenital hemihypertrophy and a complete case report." J. Pediatr., 31:403, 1947.

HEMIHYPERTROPHY SYNDROME OF THE FACE

Synonymy

S. of Curtius (I). S. de Steiner. Partial congenital hemihypertrophy of the face. Unilateral gigantism. Facial
(See first branchial arch syndromes.)

Syndromography

Clinic

Predominates in men. Present at birth, it is progressively established until skeletal maturation is achieved. The right side is affected more frequently.

There is enlargement of the ears, cheeks, half of the lips, upper jaw, jaw, zygoma and skull. Half of the tongue (deviation to the normal side and protrusion).

Generalized alveolar enlargement, early development and eruption of teeth, macrodontia, malocclusion. Vascular anomalies and skin pigmentation of the affected face may be observed. Neurological and mental alterations in 20% of cases; convulsions. It may be associated with acromegaly, normal gigantism, and hypertrophy of another part of the body.

Paraclinical examinations

Radiographic (dental) examination. The largest diameter of the canines is an early diagnostic sign.

Syndromogenesis and etiology

Possibilities of genomic alterations or environmental effects (intrauterine, incomplete twin pregnancy, hormonal and vascular).

Literature

Barwell, R.: "Case of unilateral hypertrophy of the head and face involving bones and soft parts." Trans. Path. Soc., London, 32:282-84, 1881.

Curtius, E.: "Kongenitaler, partielier Riesenwuchs mit endokrinen stoerungen." Dtsch. Arch. Klin. Med., 147:310, 1925.

Hanley, F.J.; E. Floyd, and D. Parker: "Congenital partial hemihypertrophy of the face." J. Oral Surg., 26: 136-41. 1968.

Ringrose, R.E.; J.T. Jbour and D.K. Keele: "Hemihypertrophy." Pediatrics, 36:434, 1965.

Wagner, R. In Kottmeier, H.L.: "Ueber Hemyhypertrophia und Hemiatrophia corporis totalis, nebst spontane Stremitaeten- gangren bei Sauglingen in Anschluss zu einen ungewodeniliches Fali." Acta Paediat., 20:531, 1938.

NEONATAL ADRENAL HEMORRHAGE SYNDROME

Synonymy

S. of adrenal hemorrhage in newborns.

Syndromography

Clínica

It appears with an incidence of 1% in postmortem findings in newborns. It begins between the second and seventh days of postnatal life. Occasionally it is asymptomatic and is discovered later as calcification and adrenal insufficiency.

It is accompanied by non-specific symptoms such as hyperthermia, cyanosis, tachypnea, which is why this syndrome has been called pseudopneumonia of the newborn."

There may be vomiting and convulsions, paleness, skin rash and purpura. Sometimes a painful abdominal mass can be palpated.

It often progresses to death, although regression with marked calcification is possible after 3-4 weeks.

Paraclinical examinations

Lateral abdominal radiology. It reveals a retroperitoneal mass.

Urography. Shows displacement of the kidney. Adrenal calcifications can be observed as early as 12 days after birth.

Pathology. In 70% of cases the right adrenal gland is involved: in 5-10% it is bilateral. The findings range from small hemorrhage to complete destruction of the gland.

Syndromogenesis and etiology

Stress and trauma at the time of birth, physiological involution of the adrenal glands and systemic diseases such as thrombocytopenia and syphilis are invoked.

Literature

Goldzieher, M.A. and M.D. Gordon: "Syndrome of adrenal hemorrhage in a newborn." Endocrinology, 15:165-81, 1932.

Gross, M.; P.K. Kottmeier and K. Waterhouse: "Diagnosis and treatment of neonatal adrenal hemorrhage." J. Pediat. Surg., 2:308-12, 1967.

HENNEBERT SYNDROME

Synonymy

S. of luetic otic nystagmus.

Syndromography

Clinic

Short, spontaneous attacks of vertigo and nystagmus. Digital compression of the external auditory meatus or traction of the tragus triggers spontaneous nystagmus. The tympanic membrane is intact.

Paraclinical examinations

Syphilis serology.

Syndromogenesis and etiology

Congenital syphilis.

Literature

Hennebert, C.: "Reactions vestibulaies dans les laberinthites héredo-syphitiques." Internat Arch. Laryng. Otol., 28:93-96, 1909.

HYDROXYPROLINE II SYNDROME

Synonymy

S. of hydroxyprolinemia type II.

Syndromography

Clinic

Mild mental retardation without kidney disease.

Syndromogenesis and etiology

It has been estimated that there is no metabolic phenotype for hydroxyprolinemia, but rather a problem in determination, due to the fact that hyperhydroxyprolinemia has been found in patients investigated for different abnormalities.

The basic defect is 4-hydroxyl-L-proline deficiency.

-Oxidase.

The etiology is genetic. It is transmitted as an autosomal recessive inheritance.

Literature

Berlow, S. and M. Efron: "A new cause of hyperprolinemia associated with excretion of Al pyrroline-5-carbOxylic acid." J. Pediat., 65:1122,1964.

HPERAMONIEMIA SYNDROME

Syndromography

Clinic

Elevation of ammonia in the blood produces a special clinical state in humans characterized by symptoms ranging from deterioration of psychomotor development, seizures, irritability, hypotonia, vomiting, to hepatomegaly, severe mental retardation and death.

This syndrome has an early onset in childhood. It is manifested by episodes of vomiting, screaming, agitation, followed by lethargy and stupor. Arrested growth, mental retardation, slurred speech, ataxia, eyelid ptosis.

It is important to complement the data from the patient's natural history and physical examination with laboratory studies.

Paraclinical examinations

Urine. Orotic acid. Organic acids. The latter require gas chromatography and mass spectroscopy for their detection.

Blood. Hyperammonemia (normal values: neonates up to 100 mcg/110 mL; infants and children 80 mcg/100 mL) if the blood is taken half an hour after the bottle. Elevated glutamic oxaloacetic transaminase. Cyclic neutropenia. Normal Uea. Study:

glucose, glycine and lysine.

Amino acid chromatography. Increased glycine, methylmalonic acid, citrulline, arginino-succinic acid.

Cerebrospinal fluid. Ammonia values above 480 mcg%.

Computerized axial tomography of the skull. cerebral cortical atrophy.

Syndromogenesis and etiology

Amino acids are catabolized to carbon dioxide and used in gluconeogenesis or cytogenesis, and amino groups are extracted by transamination or oxidative deamination and can be converted to ammonia.

The clinical characteristics common to states of hyperammonemia, increased glutamine in the cerebrospinal fluid, experimental evidence of the effect of high concentrations of ammonia in the brain of animals when amino acid precursors of neurotransmitters are increased, suggest that high concentrations of ammonia interfere with the metabolism of neurotransmitters.

The causes of hyperammonemia can be classified into two large groups:

Not hereditary

1. Transient neonatal hyperammonemia.
2. S. de Reye.
3. Liver damage and portocaval shunt.
4. Chemical agents: 4-pentanoic acid, valproic acid.

hereditary

1. Inborn errors of the urea cycle due to deficiencies of:
   - Carbamyl-phosphate synthetase.
   - Ornithine-transcarbamylase.
   - Argininosuccinic acid.
   - Synthetase (citrullinemia).
   - Arginino-succinase (argininosuccinic aciduria).
   - N-acetylglutamate synthetase.
2. Deficiencies of membrane transport proteins:
   - Hyperornithinemia, hyperammonemia and homocitrullinuria (HHH).
   - Lysinuric protein intolerance.
3. Secondary to other metabolic defects:
   - Organic acidurias: propionic, isovaleric, methylmalonic.
   - Pyruvate-carboxylase deficiencies.
   - Zellweger syndrome.

Literature

Friedland, G.W. and Sunshine, P. In Gellis and Kagan: Current Pediatric Therapy, 7, 1976, p. 182.

Russell, A., B. Levin; V.G. Oberholzer et al.: Hyperammonemia: a new instance of an inborn enzymatic defect of the biosynthesis of urea", Lancet. 2: 699-700, 1962.

Shih, V.E. and M.L. Efron: In Stanbury-Wyngaarden-Frederickson: Metabolic basis of inherited disease. 3 ed. McGraw-Hill, New York, 1972, p. 373.

Sunshine, P.; J.E. Lindenbaum; H.I. Levy et al.: "Hyperammonemia due to a defect in hepatic ornithine transcarbamylase." Pediatrics, 50:100, 1972.

HYPERPHOSPHATASIA SYNDROME

Synonymy

S. of macrocranium-fragile bones. S. of juvenile deforming cortical hyperostosis. S. of hereditary hyperphosphatasia. Juvenile Paget's disease.

Syndromography

Clinic

It begins in the first months of life, with fever, pain and bone fragility; poor development with dwarfism, the neck is short, the thorax is like a pigeon; delay in starting to walk, normal intelligence. Head: macroskull with prominence of mastoids and frontals due to the considerable thickening of the skull.
Trunk: short neck with keeled thorax, with everted costal ridges; biconcave vertebral bodies and kyphoscoliosis. Limbs: long bones thickened in the diaphysis and curved, particularly the femurs (in parentheses), with pelvic locking of their heads; occasionally fixed elbows; muscle weakness. Other occasional anomalies: blue sclera. cavities and tooth loss, deafness, retinopathy. Daily headaches and high blood pressure.

Paraclinical examinations

Radiology. Irregular thickening of the calvaria without sclerosis; coarse mandibular trabeculation and considerable expansion of the shafts.

Blood. Marked increase in phosphatases (alkaline and acid), leucine-aminopeptidase and uric acid.

Syndromogenesis and etiology

The lesions are located in the cortical layer, which is thickened and is composed of excess bone lamellae. The Haversian canals are occluded by an excessive fibrovascular bed: transverse defects, Milkman type.

It is transmitted by autosomal recessive inheritance. There is a late adult form.

The etiology is genetic.

Literature

Bakwin, H. and M.S. Eiger: "Fragile bones and macrocranium." J. Pediatr., 49:558. 1956.

Caffey, J.: Familial hyperphosphatasemia with ateliosis and hypermetabolism of growing membranous bone review of the clinical radiographic and chemical features. In Kaufman (Ed.): Intrinsic diseases of bones. t. 4 of "Progress in Pediatric Radiology", 1974.

Fanconi, G. et al.: "Osteochalasia desmalis familiaris. Hyperostosis deformans juvenileis, chronic idiopathic hyperphosphatasia and macrocranium." Helv. Paediat. Acta, 19:279, 1964.

HYPERGLYCINURLA SYNDROME - HYPERGLYCINEMIA

Syndromography

Clinic

It affects both sexes. From the first months of age, episodic vomiting, lethargy, and dehydration occur. Repeated episodes of infection and purpura. Mental retardation.

Physical examination. Children are small, pale, with poor muscle development.

Paraclinical examinations

Blood. Recurrent anemia, neutropenia and thrombocytopenia; hypogammaglobulinemia, ketonemia, hyperglycinemia.

Urine. Ketonuria, glycinuria.

Radiology. Osteoporosis.

Syndromogenesis and etiology

It is a defect of genetic etiology due to mutations in the mitochondrial propionylcoA carboxylase gene.

It is transmitted as an autosomal recessive inheritance, with genetic heterogeneity.

Literature

Child, B.; W.L. Nyhan; M. Borden, et a.: "Idiopathic hyperglycinemia-hyperglycinuria: a new disorder of amino acid metabolism." I. Pediatrics, 27: 522-38, 1961.

CONGENITAL ADRENAL HYPERPLASIA (CAH) SYNDROME

Synonymy

Virilizing (or feminizing) adrenal S.

Syndromography

Clinic

HAC syndromes constitute a group of diseases related to defects in different metabolic steps.

of the synthesis pathway of cortisol, aldosterone or both.

When the affected pathway is the cortisol pathway there is a compensatory increase in ACTH, if it is the mineralocorticoid pathway there is an increase in renin-angiotensin production, if the enzymatic defect affects these two pathways there is increased production of both enzymes.

The clinical manifestations will vary depending on the pathways affected and, furthermore, on the moment of synthesis where the blockage occurs and therefore will correspond to the effect on the organism of the intermediate or final defect of the products or the excess of the products due to the interruption of the metabolic pathway in question.

Taking into account this biochemical phenomenon, HACs have been classified into five groups:

Type I. It affects both females and males, but the diagnosis is suspected in males due to defects in the development of external genitalia that can range from various degrees of hypospadias to complete failure of the development of the external genitalia. The most severe norm within this type is congenital lipoid adrenal hyperplasia since there is no synthesis of steroid hormones, all newborns are phenotypically classified as girls and it presents as a severe syndrome with salt loss that produces the death of the newborn if not treated early.

Type II. The newborn presents ambiguous genitalia, there are virilization defects. Death is common in salt-wasting cases and can occur even with adequate therapy.

Type III. This is the most common form with an incidence of 1 in 5,000 live births.

In these cases there is virilization of the female external genitalia observable at birth, in the male the diagnosis is later but is manifested by rapid growth of the penis and rapid height growth. In untreated cases without salt loss, the defect produces a premature closure of the bone growth lines, eventually reaching short stature. In approximately half of the cases there is an additional defect in aldosterone synthesis that, if not adequately treated, leads to neonatal death and whose clinical manifestations may suggest pyloric stenosis.

Type IV. In this type there is clinical heterogeneity, which can range from normal external genitalia, in both the male and female sex, to highly virilized female genitalia, but this is accompanied by high blood pressure due to salt retention. However, there does not seem to be a correlation between signs of excess mineralocorticoids and the degree of virilization.

Type V. The enzymatic defect determines hypertension and hypokalemic alkalosis in these cases. In women, hypertension, primary amenorrhea and loss of secondary sexual characteristics have also been described.

Paraclinical examinations

Sexual chromatin. Determine chromatin sex.

Karyotype. Determine chromosomal sex.

Determinations of cortisol and aldosterone as well as their intermediates in blood.

Determination of HLA haplotypes.

Determinations of enzyme activity.

Molecular studies.

24 hour urine. Increased 17-ketosteroids and pregnantriol.

Blood. Low cholesterol. If there is loss of salt: hyperkalemic alkalosis.

Radiology. Breasts: hypermaturity. Bones. Stomach: to specify pyloric pseudo-obstruction.

Computerized axial tomography. It allows demonstrating adrenal enlargement.

Retropneumoperitoneum. Shows adrenal hyperplasia.

Syndromogenesis and etiology

Each type has its own characteristics with genetic etiology and autosomal recessive inheritance.

Type I. The genetic defect involves the enzyme 20,22- -desmolase that converts cholesterol to pregnenolone. The formation of pregnenolone from cholesterol presents three steps: 20-hydroxylation, 22-hydroxylation and cleavage, C20-C22 to produce pregnenolone and caproic acid. This reaction known as cholesterol cleavage is catalyzed by a specific form of cytochrome P450 called P45011A that is located in the inner membrane of the mitochondria. This gene has been cloned. Its normality in cases with lipoid HAC points to another biochemically unidentified factor in these cases of severe lipoid HAC. The genital defects presented by HAC type I seem to be related to testicular defects in the production of inducing hormones necessary for male genital organogenesis.

Type II. It is caused by a defect in the enzyme 3-beta-hydroxysteroid-dehydrogenase-isomerase (3-beta-HSD). It has been suggested the existence of at least two isoenzymes under separate genetic determinations or an enzyme that, encoded by the same structural gene, presents expression of enzymatic activity independently controlled by the testis and the adrenal gland. In both cases, syndromogenesis can be explained. This 3-beta-HSD gene is located on lp13.1.

Type III. 95% of cases depend on a defect in the 21-hydroxylase enzyme that converts 17-hydroxyprogesterone (17-OHP) into 11-deoxycortisol. The defect in cortisol synthesis increases ACTH levels and produces an increase in cortisol precursors, particularly 17-OHP proximal to the block. This determines excessive production of androgen that explains virilization. In half of the cases there is an additional defect in cholesterol synthesis (conversion of progesterone to l1-deoxycorticosterone). These two different forms of 21-hydroxylase deficiency are associated with HLA major histocompatibility complex haplotypes. The defect that only affects cortisol synthesis is associated with HLA-Bw51/5 and the severe salt-wasting form with the haplotypes HLA-(A3): Bw47;

DR7 and HLA Bw60/40. The clinical evidence of severe, moderate and mild forms combined with molecular studies carried out have allowed the analysis of multiple propositions to explain the genetic defects related to the activity of 21-hydroxylase.

Molecular studies have made it possible to locate the gene for this type of HAC at 6p21.3.

Type IV. In this type of HAC, two enzymes are involved whose individual or simultaneous deficiencies explain the clinical picture: 11-beta-hydroxylase and 18-hydroxysteroid dehydrogenase. The latter affects the end of the metabolic pathway and forms aldosterone and its isolated deficiency is accompanied by hypertension and normal genitalia. 11-beta-hydroxylase deficiency is the second cause of HAC. By in situ hybridization the gene was located at 8q21.

Type V. The enzyme deficiency has been related to the 17-hydroxylase enzyme necessary for the synthesis of cortisol and estrogen, which is why the hormones ACTH and FSH increase in the blood. The excessive synthesis of deoxycorticosterone and corticosterone produces hypertension, the loss of estrogen explains primary amenorrhea and the absence of sexual maturation.

The gene for the steroid enzyme 17-hydroxylase appears to be located on chromosome ID.

Literature

Bongiovanni, A. M. In Stanbury-Wyngaarden-Frederickson: Metabolic basis of inherited disease. 3 ed. Ed. McGraw-Hill New York, 1973.

McKusick, V.A.: Mendilian inheritance in man, 10th ed. v. 2. The Johns Hopkins University Press. Baltimore 1992, pp. 1188- -1198.

Marchand, F.: Ueber allgemeine Hyperplasie der Nebennieren bei Pseudohermaphrodytismus femeninus. Feettscr. Virchov. t. I, 554, 1897.

CONGENITAL ADRENAL HYPERPLASIA SYNDROME TYPE I

Synonymy

S. de Prader-Gurtner. Desmolase deficiency S. 20-21.S. of adrenal lipoid hypertrophy.

Syndromography

Clinic

It begins in the neonatal period with anorexia, vomiting, strange skin color ("honey skin"), and weight loss that is not recovered. Apathy, and diarrhea with dehydration.

Physical examination. Normal genitalia in females or male pseudohermaphrodites (testes), with abdominal cryptorchidism sometimes.

Paraclinical examinations

Urine. Does not contain 17-ketosteroids, 17-ohketosteroids or pregnancyriol.

Blood. Hyponatremia and hyperkalemia.

Karyotype. Male with external pseudohermaphroditism, or female with normal female genitalia.

Syndromogenesis and etiology

This syndrome is due to a deficiency of desmolase 20-21 (which converts cholesterol into pregnenolone) and therefore testosterone, androgens, and aldosterone are not synthesized, which produces total adrenal insufficiency due to which newborns die between 1-8 months due to infections.

The etiology is genetic and is transmitted by autosomal recessive inheritance, with frequent consanguinity of the parents.

Literature

Brutschy, P.: “Hochgradige Lipohyperplasie beider Nebennieren mit herdfbermigen Kalkablagerungen bei einem Fall von Hypospadia peniscritalis mit unechterakzessorischer Nebeniere am rechten Hoden.” Frankfurt. Z. Path., 24:203, 1921.

O'Dohert, NJ.: "Lipoid adrenal hyperplasia." Guy Hosp. Rep. 113:368, 1964 (cited. In Hubble Pediatric Endocrinology). Ed. Blackwell, Oxford, 1969. p 261.

Prader, A. and H.P. Gurtner: *Das Syndrome der Pseudo-hermaphrodistismus masculinus bei kogenitaler Nebennierenn der Hyperplasie ohne Androgenueberpro-duktion". elv. Paediat. Acta, 10:397,1955.

CONGENITAL ADRENAL HYPERPLASIA SYNDROME TYPE II

Synonymy

S. de Bongiovanni. S. of 3 beta-dehydrogenase deficiency.

Syndromography

Clinic

It begins a few days or weeks after birth.

Physical examination. Males: incompletely developed genitals with varying degrees of hypospadias, or complete lack of masculinization with a vagina. Females: moderate virilization, with hypertrophy of the clitoris, fusion of the large labia, without displacement of the urethral orifice. Development is inhibited and does not respond to replacement treatment.

Paraclinical examinations

Laboratory exams. Low testosterone and high production of dehydroepiandrosterone (DHEA).

Syndromogenesis and etiology

Congenital adrenal hypertrophy with 3-beta- -hydroxysteroid -dehydrogenase deficiency resulting in defective testosterone production and less androgenic activity. A small peripheral partial transformation of DHEA into testosterone explains the lower virilization in females and males.

Of genetic etiology, the altered gene affects both the adrenal cortex and the male gonad.

Literature

Bongiovanni, A.M.: "Unusual steroid pattern in congenital adrenal hyperplasia.: Deficiency of 3 beta-hydroxyldehydrogenase." J. Clin. Endocrinol., 21:860, 1961.

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CONGENITAL ADRENAL HYPERPLASIA SYNDROME TYPE II

Synonymy

S. de Debré-Fibiger. S. of 21-hydroxylase deficiency.

Syndromography

Clinic

It occurs in females at birth and in males around 2-3 years of age.

Physical examination. In females: virilization with clitoral hyperplasia and varying degrees of labio-scrotal fusion, with normal uterus and ovaries, urogenital sinus. In men: normal for the first two years and then progressive enlargement of the penis (macrogenitosomia) with pubic and axillary hair, acne and change in voice. Excessive somatic development, with muscular habit, which simulates a small adult. Bone hypermaturity, at 5 years old for example, they have a size of 8; early epiphyseal fusion that ultimately results in short stature or dwarfism. Females that are not treated show a viriloid appearance and then baldness.

When the syndrome coexists, salt loss (30%), in the first days or weeks there is repeated explosive vomiting, without pyloric tumor (pseudopylorostenosis), dehydration, anorexia and shock, with possible sudden death.

Paraclinical examinations

Urine. Increase in 17-ketosteroids and pregnantriol (more than 1 mg/24 h). The 11-oxygenation test can be done only with urine from a void and gives values greater than 0.62.

Syndromogenesis and etiology

It is the most common form of congenital adrenal hyperplasia (90%). Deficiency of 21-hydrolase causes large production of androsterone, which is peripherally converted to testosterone, with normal cortisol and occasional aldosterone deficiency.

It is transmitted by autosomal recessive inheritance, affecting several siblings without distinction of sex.

The etiology is genetic.

Literature

Bartter, F.C.: A.P. Forbes and A. Leaf: "Congenital adrenal hyperplasia associated with the adreno-genital syndrome: an attempt to correct its disordered hormonal pattern." J. Clin. Invest., 29:797, 1950.

Bongiovanni, A.M.: In Stanbury-Wyngaarden-Frederickson: Metabolic basis of inherited disease. 3 ed. Ed. McGraw-Hill, New York. 1972, p. 875.

Debré, R. and G Semelaigne: "Considerable hypertrophy des capsules surrenales chez un nourrison mort a 10 mois, sans avoir augmente de poids depuis la naissance." Bull. Soc. Pediat. Paris, 23:270, 1925.

CONGENITAL ADRENAL HYPERPLASIA SYNDROME TYPE IV

Synonymy

S. of Eberlein-Bongiovanni. S. of 11-hydroxylase deficiency.

Syndromography

Clínic

It is characterized by an adrenal virilizing syndrome and arterial hypertension. It constitutes 5% of all cases of congenital adrenal hyperplasia.

Paraclinical examinations

Blood. It differs from type III in that there is accumulation of DOCA in this medium.

Urine. Low pregnancyriol, and increased 17-ketosteroids and androsterone.

Syndromogenesis and etiology

Deficiency of 11-hydroxylase prevents the formation of cortisol and DOCA accumulates in the blood, causing hypertension.

Literature

Dyrenfurth, I.; S. Sybulski, et al.: "Urinary corticosteroid excretion patterns in patients with adrenocortical dysfunction." J. Clin. Endocr.. 18:391. 1958.

Eberlein, W.R. and A.M. Bongiovanni: In Stanbury-Wyngaarden- -Frederickson: Metabolic basis of inherited disease. 3 ed. Ed. McGraw-Hill, New York, 1972. p. 873.

CONGENITAL ADRENAL HYPERPLASIA SYNDROME TYPE V

Synonymy

S. de Biglieri. S. of 17-hydroxylase deficiency.

Syndromography

Clinic

It is negligible in the first years, except in men who have hypospadias. In both sexes the phenotype is female.
During childhood, respiratory infections occur repeatedly.

Physical examination. The genitals are normal in the female, in the male there is hypospadias and cryptorchidism. Puberty is inapparent upon reaching that age, absence of menarche and secondary sexual characteristics. Moderate hypertension since puberty. In adulthood there is hypokalemic alkalosis.

Paraclinical examinations

Urine. Lack of 17-ketosteroids.

Syndromogenesis and etiology

It is produced by a 17-hydroxylase deficiency, which reduces the formation of cortisol and this causes an increase in ACTH, which in turn induces hyperproduction of DOCA with consecutive hypertension; On the other hand, the formation of androgens and estrogens (causing the absence of puberty) is minimal. The hyperproduction of aldosterone that occurs in adulthood causes hypokalemic alkalosis.

Literature

Biglieri, E.G.; M.A. Herrow and N. Brosto: *17 Hydroxylation deficiency in man". J. Clin. Invest., 45:1946, 1966.
New, M.: "Male pseudohermaphrodytism due to 17 hydroxylation deficiency" J. Clin. Invest., 49:1930, 1970.

GINGIVAL HYPERPLASIA SYNDROME -HIRSUTISM-CONVULSIONS

Synonymy

S. of gingial fibromatosis-epilepsy-hypertrichosis-mental retardation. S. of gingival fibromatosis-hypertrichosis.

Syndromography

Clinic

Sometimes the newborn already has hirsutism, other times it occurs during puberty. Facies: coarse, with hirsutism and lip stubble, sometimes acromegaloid. Hypertrichosis: generalized, affects hair that is black and abundant: eyebrows are thick and black. Hypertrichosis extends to the back, sacral region, genitals, arms and legs. Gums: thickened, can cover the teeth.

Epilepsy, centrocephalic type. Mental retardation, delay in walking and talking, IQ may be low (59%).
Other occasional anomalies: narrow second cervical vertebra with long odontoid process, anomalous first rib; Sometimes auricular, nasal and dental alterations are associated, constituting the so-called Zimmerman-Laband syndrome.

Paraclinical examinations.

Gingival biopsy. Connective hyperplasia of gums and collagen fibers.

Syndromogenesis and etiology

Many cases are due to the ingestion of hydantoin by the mother during pregnancy, but another large group does not have this history.

The hypertrophy of the gums, which are firm and have a smooth surface, are due to collagenous tissue covered by stratified epithelium.

It is transmitted in an autosomal dominant manner; genetic heterogeneity (sometimes recessive)

The etiology is genetic.

Literature

Goodman, R.M. and R.J. Gorlin: The face in genetic disorders. Ed. Mosby, St. Louis, 2 ed., 1977. p. 329.
Snyder, C.H.: "Syndrome of gingival hyperplasia hirsutism and seizures." J. Pediat., 67:499, 1965.

HYPERTELORISM-HYPOSPADIA SYNDROME

Synonymy

S. of telecanth with associated malformations.

Syndromography

Clínica

Patients who suffer from this syndrome appear hypertelorism, but the orbits are not distanced, only the inner canthus of the eyes (telecanthus or dystopia cantorum).
Hypospadias manifests in males. Medium or minimal mental retardation. Other probable anomalies: cryptorchidism, brachycephaly, congenital heart disease, dental anomalies, and diastasis recti abdominis.

Syndromogenesis and etiology

It is transmitted in an autosomal dominant manner, with greater penetrance in males. The females of the family only show telesinging.

The etiology is genetic.

Literature

Christian, J.C.; D. Bixler; D.C. Blythe et al.: Familial telecanthus with associated congenital anomalies". Birth defects: orig. Series, vol. II:82, 1969.
Michael, E. and W. Mortier: "Association of hypertelorism and hypospadias." Helv. Paediat. Acta, 27:575, 1972.
Opitz, J.M.; R.L. Sumit and D.W. Smith: "The 888 syndrome familial telecanthus with associated congenital anomalies." Birth defects: original Series. vol. II,: 86, 1969.

HYPOCHONDROPLASIA SYNDROME

Synonymy

S. of chondrohypoplasia.

Syndromography

Clinic

This bone dysplasia is not detectable at birth, size defects begin around 3 years of age, final height is 46.5-60 inches. With weight, arching of the legs and genus varum occur. The patients present lumbar lordosis, there is no facial dysmorphism. Occasionally, frontal bulging or brachycephaly may occur. Chubby hands and feet. Limitation of elbow extension and supination. Mental deficiency is rare.

Paraclinical examinations

Radiology (bone survey). Short tubular bones with slightly irregular metaphyses, longer fibula. Shortening of the lumbar pedicles, narrowing of the spinal canal.
Pelvis square, iliac wings short.

Syndromogenesis and etiology

The etiology is genetic with autosomal dominant inheritance. It has been suggested that some cases of hypochondroplasia are caused by a defect in insulin-like growth factor I (1G FI). Strong linkage of the IG FI locus and a group of hypochondroplastic patients has been demonstrated. It has also been suggested that the genes for achondroplasia and hypochondroplasia are alleles.

Literature

Mullis, P.E. et al.: "Growth characteristics and response to growth hormone therapy in patients with hypochondroplasia: genetic linkage of the insulin-like growth factor I gene at chromosome 12q23 to the disease in a subgroup of these patients." Clin. Endocr., 4:265-274, 1991.

TRANSIENT NEONATAL HYPOGLYCEMIA SYNDROME

Synonymy

Symptomatic transient neonatal hypoglycemia. S. of transient neonatal hypoglycemia.

Syndromography

Clinic

Prevalence in males born to mothers who have been reported to have had preeclampsia. This is a normal newborn with a history of crying and spontaneous breathing whose symptoms begin 2 hours to 7 days after birth, presenting tremor, cyanosis, seizures, periods of apnea or respiratory distress, rotating eye movements, apathy, flaccidity, weak or high-pitched crying, and difficulty feeding.

Physical examination. It is frequently a low-weight newborn, below the 10th mark. percentile.

When it occurs in twin newborns, it generally affects the one with the lowest weight.

There is a rapid response to glucose administration.

Paraclinical examinations

Blood. Hypoglycemia, generally low calcium, polycythemia.

Cerebrospinal fluid. Normal, except low glucorachia.

Syndromogenesis and etiology

It has been related to possible malnutrition in intrauterine development, primary brain defect or failure in the glycogenolysis mechanism. It is proposed that during intrauterine life the fetus receives glucose through the placenta in a way that does not depend on its glycogen reserves or its glycogenolysis mechanisms. After childbirth, the few glycogen reserves, especially in small and premature infants, are quickly consumed, there is also high glucose needs by the brain and on the other hand there is immaturity of the enzymes of gluconeogenesis so glucose production is limited. There may also be temporary hyperinsulinism that acts as a mechanism that produces transient hypoglycemia.

There are some factors that can aggravate this condition, such as fetal distress due to asphyxia, hypothermia or taxemia.

The etiology is unknown.

Literature

Cornblath, M.; GB. Odell and E.Y. Levin: “Symptomatic neonatal hypoglycemia associated with toxemia of pregnancy.” J. Pediat., 55:545-62, 1959.

Cornblath, M., and S.H. Reisner: “Blood glucose in neonate and its clinical significance.” New Eng. J. Med., 273:378-81, 1965.

Pediatric Clinics of North America. Pediatric and Adolescent Endocrinology. Interamerican Publishing House. v. 4, p. 1033, 1987.

SEX-LINKED ISOLATED RECESSIVE INFANTILIAL HYPOPARATHYROIDISM SYNDROME

Syndromography

Clinic

It begins in males during the first year of life. There is an absence of monilial infections and Addison's manifestations.

Paraclinical examinations

Blood. Hypocalcemia with hyperphosphoremia.

Radiology. Absence of signs of rickets or osteomalacia

Syndromogenesis and etiology

Of genetic etiology and recessive inheritance linked to the X chromosome. The gene is located in Xq26-q27.

The syndrome appears to be due to parathyroid agenesis.

Literature

Currarino, G.: "Hypophosphatasia": In Kaufmann, H.J. (Ed.): Progress in Pediatric Radiology, v.4, Basel Karger, 1973, p.469.

MacPherson, R.I. et al.: "Hypophosphatasia in an adult." J. Bone Joint Surg., 52:1477, 1970.

Peden, V.H.: "True idiopathic hypoparathyroidism as a sex-linked recessive trait." Amer. J. Human Genet., 12:323-37, 1960.

Rathbun, J.C.: "Hypophosphatasia: new developmental anomaly." Am. J. Dis. Child., 75:822, 1948.

Taiz, L.S.: C. Zarate-Salvadore and E. Schwartz: "Congenital absence of parathyroid and thymus in an infant." Pediatrics, 38:412-18, 1966.

Thakker, R.V. et al.: "Mapping of the X-linked idiopathic hypoparathyroid gene to Xq26-Xq27 hy linkage studies (abstract)." Cytogenet. Cell Genet. , 51:1089, 1989.

FAMILIAR HYPOTHYROIDISM SYNDROMES

Synonymy

S. of dyshormonogenesis. Inborn errors of thyroxine metabolism.

Syndromography

Clínic

Goiter is sometimes observed in the neonatal period, but more often it is detected at school age. It is manifested by diffuse, discrete goiter, without nodules, which may go unnoticed by the child's relatives; it usually precedes the functional manifestations. The symptoms of hypothyroidism are mild: minimal myxedema of the face and neck and dry skin and hair. The size is low due to short legs and sometimes

They present epiphyseal dysgenesis (elbows, ankles or hips), after having grown well for several years. Moderate school delay with bradypsychia. Inferior spastic paresthesia, sometimes, which in areas without goiter endemic suggests dyshor- monogenesis.

Paraclinical examinations

Blood. The uptake of radioactive iodine and its retention at high values during the first two hours, as well as the discharge effect of glandular iodine by perchlorate, contribute to the diagnosis.

Thyroid biopsy. Carry out histological and biochemical study.

Syndromogenesis and etiology

In reality, these are several hypothyroid syndromes, which in common present various enzyme deficiencies (some still unknown) that hinder the formation of thyroxine, which in turn produces an increase in pituitary TSH, which causes thyroid hyperplasia and, depending on the intensity of the hormonal deficiency, causes goiter plus hypothyroidism or euthyroid goiter.

Several ways have been described:

1. Iodine trapping defect. Of the 14 cases described with this syndrome, 8 were in Japan. There is goiter but, in contrast to the other defects, the uptake of radioactive iodine is low. The salivary glands and stomach also have a lack of ability to concentrate iodide. The biochemical defect is unknown but iodidepermease deficiency is a possibility. Goiter and hypothyroidism usually appear in the first months of

life.

2. Iodide organization defect. After iodide is trapped by the thyroid, it is rapidly oxidized by H,0, and thyroid peroxidase and binds to tyrosine; But in this defect the iodide is not incorporated into the metabolic pathway and can be rapidly released from the thyroid by administration of perchlorate.

Three types of organizational defects have been described:

Complete absence of peroxidase activity in a severe clinical form of goitrogenic hypothyroidism.

Failure of binding between the prosthetic group of hematin and thyroid apoperoxidase in euthyroid patients with goiters.

- Inactive peroxidase due to an alteration in the complex once formed.

Deficient organization can be seen in Pendred syndrome, in which the biochemical defect remains unknown although peroxidase activity is normal.

In the clinical aspect, perceptual deafness is added, which facilitates its recognition.

3. Coupling defect. After iodine binds to tyrosine in thyroglobulin and forms iodotyrosine, an intramolecular rearrangement occurs, leading to coupling of iodotyrosines and forming diiodothyronines. The biochemical alterations involved in this reaction are poorly understood. Alterations in the coupling enzymes or an abnormality in their configuration have been suggested.
4. Defect of thyroglobulin synthesis. Patients with this disorder release iodinated proteins and calorically inactive polypeptides from the thyroid into the blood.
   Since the synthesis of thyroglobulins is very complex, this condition possibly has diverse etiologies. In some cases there is inadequate protein iodination and poor thyroxine biosynthesis. There is also a high rate of production of iodohistidine, which is not deiodinated, but is excreted in the urine.
5. Deiodinase or dehalogenase deficiency. In these cases, the deionization of thyroid iodine fails; the labeled monoiodothyroxines and diiodothyroxines pass into the blood and are lost through the urine: in some cases the deficiency is associated with Werdnig-Hoffmann syndrome.
   The etiology is genetic. It is transmitted in an autosomal recessive manner, with frequent consanguinity of the parents and female predominance.

Literature

Hamilton, J.C.; M.H. Soley; W.A. Reiley et al.: “Radioactive iodine studies in childhood hypothyroidism.” Am. J. Dis. Child., 66:495, 1943.

Hutchinson, J.H. In Gardner: Endocrine and genetic diseases of childhood. Ed. Saunders, Philadelphia, 1969, p.253.

Osler, W.: "Internal secretion considered in their Physiologic Pathological and Clinical Aspects." Trans. Cong. A. Phys. Surg., 4:169, 1897.

Stanbury, J.B. In Stanbury-Wyngaarden-Frederickson: The metabolic basis of inherited disease. 3 ed. McGraw-Hill, New York, 1972, p. 223.

HOLTERMÜELLER-WIEDEMANN SYNDROME

Synonymy

S. of skull in clover. Skull S. from Kleeblattschadel- -Trefoil.

Syndromography

Clinic

The cloverleaf skull has a trilobular configuration.

from light to accentuated and in some cases asymmetrical.

There is downward displacement of the ears. Hypoplasia of the upper jaw and relative prognathism of the mandible.

Sunken nasal root and beak-shaped nasal dorsum. Significant proptosis, ocular hypertelorism, antimongoloid palpebral fissures and exophthalmia, all of the above give it a grotesque appearance.

There may be hydrocephalus and psychomotor and mental retardation.

Malformations such as facial clefts, absence of external auditory canal and congenital heart disease appear less frequently.

In some cases it is associated with bone deformities (achrondroplasia), but ankylosis of the elbows is rare.

Paraclinical examinations

Radiology. Skull with trilobular contour, increased digital impressions and vault with thin and distorted walls with a honeycomb appearance. There may be synostosis of the coronal, lambdoid, and metopic sutures with bulging of the brain through the open sagittal suture.

Synostosis and shortening of the skull base, small posterior skull fossa, poor development of the sphenoid and ethmoid bones.

Syndromogenesis and etiology

It is proposed that the cloverleaf skull is a sign rather than a specific condition; it may constitute an isolated anomaly without a known etiology or basic defect or may accompany others.

syndromes with craniosynostosis such as Apert, Pfiffer, Crouzon, or at low stature of prenatal onset such as thanatophoric dysplasia.

The abnormality in ossification translates a synostosis of the skull sutures in the intrauterine stage, which results in early hydrocephalus.

This type of skull is seen in many craniosynostosis syndromes.

Literature

Angle, C.R.: M.S. Mcintire and R.C. Moore: “Cloverleaf skull: "Kleeblattschadel deformity syndrome". Amer. J. Dis. Child., 114:198-202, 1967.

Cohen, M.M. Jr: “An etiology and nosology overview of craniosynostosis syndrome". Birth Defects: Orig. Art. Ser. XI (2): 137-189, 1973.

Iannaccone, G. and G. Gerlini: "The so-called cloverleaf of skull syndrome". Pediatr. Radio.. 2:175-183, 1974.

Holtermüeller, K. and H.R. Wiedemann: “Kleeblattschadel syndrome" . Med. Wschr., 14:439-46, 1960.

HUNTER SYNDROME

Synonymy

S. of mucopolysaccharidosis type II.

Syndromography

Clinic

This degenerative, often fatal disorder affects one in every 100,000 births.

The clinical characteristics of this syndrome are progressively established. Growth deficiency becomes apparent between the first and fourth years of life. The facies is coarse and does not differ much from Hurler syndrome, it also presents thick and semi-flexed joints. Hepatosplenomegaly, thickened skin, hypertrichosis, hernias, progressive deafness, hoarse voice, delayed tooth eruption.

It differs from the Hurler in that:

1 The corneas are clear.

2. Lagiba is less marked.

3. The installation of the clinical picture is more gradual.

4. It does not affect the female sex.

Physical examination. Dwarfism, grotesque facies, joint rigidity. Nodular lesions on the skin of the back and arms, hepatosplenomegaly. Absence of hump, absence of opacity of the cotnea. In some cases there is atypical retinitis pigmentosa.

Paraclinical examinations

Radiology (bone survey). Signs of bone dysostosis similar to Hurler syndrome are observed.

Urine metabolic tests. Tolouidine blue positive. Mucopolysaccharide chromatography excretion of dermatansulfate and

Enzyme activity studies in serum, leukocytes and fibroblasts. Decrease or absence of iduronate-sulfatase.

Syndromogenesis and etiology

It is a disease of genetic etiology determined by a mutation on the X chromosome in the Xq25-q27 region that affects the normal function of the enzyme iduronate sulfatase, essential for the fragmentation of two mucopolysaccharides: dermatan sulfate and heparan sulfate, which causes the cytoplasmic accumulation of mucopolysaccharides, which explains the clinical symptoms of the disease. It belongs to the group of lysosomal conditions.

It is transmitted by sex-linked autosomal recessive inheritance.

V is observed in men.

The etiology is genetic.

Literature

Hunter, C.: “A rare disease in two brothers". Proc. Roy. Soc. Med., 10:104-16, 1917.

Leray, J.G. and A.C, Crocker: "Clinical definition of the Hurler-Hunter phenotypes. A review of 50 patients." Am. J. Dis. Child., 112:518-530, 1966.

Sjoberg, I. et al.: Hunter's syndrome: A deficiency of L-idurono-sulfate sulfatase". Biochem. Biophys. Res. Commun. 54:1125, 1973.

Upadhyaya, M. et al.: "Localization of the gene for Hunter syndrome on the long arm of X chromosome." Hum. Genet., 74:391-398, 1986.

Williams, H.E.: "Heritable disorders of mucopolysaccharide metabolism." Qual. Med. 106:306-11, 1967.

HURLER SYNDROME

Synonymy

S. of mucopolysaccharidosis type I. S. of lipochondrodystrophy (improper). S. de Pfaundler-Hurler. S. of dysostosis multiplex. S. of gargoyle.

Syndromography

Clinic

It affects both sexes. The clinical onset is in childhood.

After a few months of normal growth, the physical and mental faculties show progressive deterioration.

It is characterized by severe mental retardation with skeletal anomalies: dwarfism, gargolic facies (enlarged skull with synostosis of the sagittal suture, wide and depressed nasal root, prominent jaw). Wide hand with short fingers, flexion contracture of the knees and elbows.

Sometimes deafness, opacity of the conea, hepatosplenomegaly, prominent abdomen, hernia. It can be associated with a wide variety of heart valve diseases, large arteries, coronary arteries, as well as

Physical examination. Hydrocephalus, grotesque facies, prominent nose and ears, thick lips, hypertelorism. Macroglossia, thoracolumbar kyphosis, joint stiffness, thoracic deformity, dwarfism, corneal opacity and retinal degeneration, hepatosplenomegaly, thick skin with nodules in the scapular region, hirsutism. Short neck, short thick toes, coxa valga, genu valgum, club foot.

Paraclinical examinations

Blood. Reilley-Alder body (metachromatic inclusion) in polynuclear cells. Bone marrow: Reilley bodies in histiocytes and lymphocytes.

Radiology. Enlargement of the axis of long bones, vertebral convexity, posterior displacement of one or two of the first vertebral bodies, coxa valga. Thickening of the cranial vault; there may be absence of frontal sinuses.

Lateralization of jaws and teeth, dental cysts.

Increase in size of the sella turcica that presents a characteristic ' shoe' shape. Widening and shortening of

long bones and hypoplasia and spur formation in the vertebrae of the lumbar spine.

Urine. Accumulation in tissue and excretion in urine of dermatan and heparan sulfate.

Liver biopsy. Kupffer cells and other macrophages enlarged and filled with metachromatic granular material.

It can progress late to cirrhosis.

Syndromogenesis and etiology

Mucopolysaccharidosis that is inherited with an autosomal recessive trait. It appears to be due to a lack of alpha-1-iduronidase activity with exaggerated deposition of mucopolysaccharides (chondrointin sulfate [dermatansulfatol) and heparin sulfate (heparan sulfate) in many tissues.

Literature

Hers, H.G. and E. Van Hoof: Lysosomes and storage diseases. Academis, New York, 1973.

Hurler, G.: "Ueber einen Typ multipler Abartungen, vorwiegend am Skelettsystem. Zschr. Kinderh., 24:220, 234, 1919.

Stambury, J.B. et al.: The metabolic basis of inherited diseases. 5th ed. McGraw-Hill, New York, 1983.

Williams, H.E.: "Heritable disorders of mucopolysaccharide metabolism", Calif. Med., 106:306-31 1, 1967.

HURLER-SCHEIE SYNDROME

Synonymy

S. Hurler-Scheie compound. IH/S mucopolysaccharidosis S.

Syndromography

Clínic

A gradual development of signs and symptoms is observed during the first year of life, especially between 1 and 2 years. During the first year, growth tends to be accelerated and then it stops and finally there is growth deficiency. They develop macrocephaly, depressed nasal bridge, thick lips, corneal opacity, micrognathia, thick skin with hirsutism, moderate joint limitation, umbilical or inguinal hernia, hepatosple-nomegaly, deafness, arachnoid cysts. Respiratory infections, rhinorrhea, otitis.

In general, the clinical manifestations are less severe than in Hurler syndrome but very similar.

Paraclinical examinations

Radiology (bone survey). Moderate dysostosis.

Urine metabolic tests. Chromatographic detection of dermatan sulfate and heparan sulfate mucopolysaccharides. Toluidine blue

enzymatic activity. Alpha -L-iduronidase defect.

Syndromogenesis and etiology

It presents autosomal recessive inheritance, the mutant gene for Scheie syndrome and Hurler syndrome seem to be allelic mutations that affect the enzyme alpha-L-iduronidase as a basic defect, causing storage of mucopolysaccharides in parenchyma and mesenchymal tissue.

Literature

Stevenson, R.E. et al.: "The iduronidase-deficient mucopoly-saccharidosis: clinical and roentgenographic features." Pediatrics, 57:111-118, 1976.

HUTCHINSON SYNDROME

Synonymy

S. of Hutchinson's triad.

Syndromography

Clinic

The triad is made up of: 1. Hutchinson's teeth: dirty-colored teeth, with a large separation between them. Upper incisors narrow, small and somewhat arched on the sides, with a depression on the cutting edge. The lower incisors are pointed. Poorly developed first molar. 2. Deafness. 3. Interstitial keratitis.

Paraclinical examinations

Examination to determine hearing acuity.

Syndromogenesis and etiology

These symptoms are congenital and develop during childhood and the first years of life in cases of congenital syphilis. They can also be observed in rickets and traumatic injuries.

Literature

Hutchinson, J.: "On the different forms of inflammation of the eye consequent on inherited syphilis." Ophth. Hosp. Rep., 1:191-203, 1858, 226-244, 1858; 2:54-105, 1859.

——— Medical Classics, 5:138-46, 1940.

HUTCHINSON-GILFORD SYNDROME

Synonymy

S. of progeria. S. of premature senility. S. of Gilford.

Syndromography

Clínic

Patients have a normal appearance at birth in both sexes and during the first year of life. Afterwards they begin to lose weight, delayed growth is observed (height and weight below the third percentile). Hair loss leading to baldness, absence of eyelashes, loss of subcutaneous fat, dwarfish appearance, small face, prominent eyes, pointed nose, irregular teeth and everything gives the child a senile appearance.

Brown patches are observed on the skin. Infantile sexual organs, protrusion of the abdomen and poor muscle development. The veins are very visible on the head due to the absence of subcutaneous fat, which reinforces the senile appearance.

Early opacity of the lens. Generalized atheromatosis.

Tendency to cerebral or cardiac vascular ischemia.

Limitation of joint motility due to contractures and anchi-losis.

The appearance is so characteristic that it has been rightly said that nothing is more similar to progeria than another case of progeria (Dieguez).

An incomplete form of this condition occurs in Souquez-Charcot syndrome; A limited form is Gottron syndrome and another variant is Bamatter-Franceschetti-Klein-Sierro.

Death occurs in the first or second decade of life.

Paraclinical examinations

Blood. Increased lipids. Hypercholesterolemia.

Urine. Aminoaciduria

Radiology. Premature fusion of the epiphyses, thinning of the long bones with decalcification and osteoporosis.

Chromosome patterns. Normal.

Syndromogenesis and etiology

It has been proposed: polyglandular insufficiency, adrenal insufficiency, diencephalon-pituitary disorders, or complex mesoectodermal

Recently it has been related to an alteration in the growth and regenerative capacity of fibroblasts. In some cases these alterations are present even if their parents are clinically healthy (heterozygous).

Literature

Hutchinson, J.: "Congenital abscense of hair and mammary glands with atrophic conditions of skin and its appendages." Med. Chir. Trnas., 69:473-477, 1886.

Franklyn, P.P.: “Progeria in siblings", Clin. Radiol. (Edimb.), 27:327-333, 1976.

FAMILIAR AMAUROTIC DEIDIOCIA SYNDROMES

Synonymy

S. of lipoidosis due to gangliosides.

Several syndromes are considered, including: Tay-Sachs syndrome and Bielschowsky syndrome. Although with similar histological alterations, these syndromes can be considered as different clinical entities due to their different genetic transmission and the quantitative and qualitative differences of the gangliosides involved.

Literature

Stanbury, J.B.; J.B. Wyngaarden and D.S. Frederickson: The metabolic basis of inherited disease. ed. 2, McGraw-Hill, New York, 1966.

COMBINED IMMUNODEFICIENCY-ADA DEFICIT SYNDROME

Synonymy

S. of adenosine deaminase (ADA) deficiency.

Syndromography

Clinic

This syndrome was described by Biblet in 1972, in an infant who presented severe immunodeficiency associated with an absence of ADA in his red blood cells.

It begins in the first months with repeated infections and by a year, a deterioration of B cells and immunological deficiency is seen.

Paraclinical examinations

Blood. Marked lymphopenia.

Immunological study. Deficit in cell-mediated immunity and depression of delayed hypersensitivity; with

As age progresses, the functions of T and B cells disappear.

Radiology. Wide pelvis with horizontal acetabular roof, small sacrociatic notch. Thoracolumbar platyspondyly; anterior costal ends, short, wide and concave.

Syndromogenesis and etiology

The deficiency or absence of the enzyme adenosine deaminase causes severe immunodeficiency due to the accumulation of adenosine, which is toxic to lymphocytes.

It is transmitted in an autosomal recessive manner. Its frequency is minimal, about thirteen cases have been diagnosed.

The etiology is genetic.

Literature

Dissing, J. and B. Knudsen: “Adenosine deaminase deficiency and combined Immunodeficiency syndrome.” Lancet, 2:1316, 1972.

Giblett, E.R.; J.E. Anderson; E. Cohe et al.: "Adenosine deaminase deficiency in two patients with severely impaired cellular immunity." Lancet, 2:1067, 1972.

Seakins and Harkness: Inborn errors of immunity and phagocytosis. Lancaster, MPT. 1979.

HEREDITARY FRUCTOSE INTOLERANCE SYNDROME

Synonymy

S. of fructose intolerance. S. of fructosemia.

Syndromnography

Clinic

No symptoms until the patient eats foods containing fructose. The first symptoms appear in childhood, are observed when sucrose or fructose are added to the diet and are characterized by hypoglycemic attacks related to the ingestion of these foods; occur: vomiting, sweating, unconsciousness, convulsions, despondency until reaching the

Coma.

Difficulty in development, cachexia that can lead to

death.

In older children and adults, the strong aversion to fruits and sweets prevents other manifestations.

Physical examination. Jaundice, hepatomegaly and sometimes splenomegaly, hemorrhagic diathesis, ascites. Poor development without affecting intelligence.

Paraclinical examinations

Blood. Hypoglycemia following fructose ingestion. Transaminases and 1-6-diphosphate -aldolase increased, inorganic serum phosphorus decreased. Hyperbilirubinemia. Intravenous fructose tolerance test.

Urine. Albuminuria, aminoaciduria, fructosuria (after ingestion of fructose).

Syndromogenesis and etiology

It is due to the congenital absence of aldolase-1-phosphate-fructose which causes accumulation of fructose-1-phosphate in the liver, which leads to liver cirrhosis unless all foods containing fructose are avoided.

At least three different defects of the aldolase B gene have been described, indicating allelic heterogeneity of the defect.

The etiology is genetic and is transmitted in an autosomal recessive manner.

Literature

Cross, N.C.P. et al.: "Molecular analysis of aldolase B genes in hereditary fructose intolerance." Lancet, 335:306-309, 1990.

Chambers, R.A. and R.T.C. Pratt: "idiosyncrasy to fructose." Lancet, 2:340, 1956.

Froesch, E.R.; A. Prader; A. Labhardt et al.: "Die hereditare Fructose intoleranz, eine bisher nicht bekannie kogenitale Stoffwechsel Storung". Sweiz. Med. Wschr., 86: 1 168-71, 1957.

Froesch, E.R.; A. Prader: H.P. Wolf, et al.: Die Hereditaere Fruktoseintoleranz". Helv. Paediat. Acta, 14:99, 1959.

Froesch, E.R. In Stanbury, Wyngaarden and Frederickson: The metabolic basis of inherited disease. 3 ed. McGraw-Hill, New York, 1972. p.131.

Sachs, B.; L. Sternfeld and G. Kraus: “Essential fructosuria its a pathophysiology". Am. J. Dis. Child., 63:252, 1942.

LYSINE INTOLERANCE SYNDROME

Synonymy

S. of familial protein intolerance.

Syndromography

Clinic

It affects the patient from birth. It is manifested by episodes of vomiting, abdominal pain, diarrhea and coma; severe motor retardation, hepatosplenomegaly, protein aversion, abnormal delayed intelligence.

Paraclinical examinations

Blood. Elevated plasma levels of lysine and arginine. During the attacks marked ammonemia.

Syndromogenesis and etiology

Inability to degrade lysine. This amino acid inhibits arginase and interferes with urea synthesis, leading to ammonia poisoning.

It has been shown to be a defect in L-lysine: NAD-oxidoreductase in the liver. It appears to be the same defect as hyperlysinemia.

Genetic etiology and possible autosomal recessive inheritance.

Literature

Colombo, J.P; R. Richterich; D.A. Spahr et al.: "Congenital lysine intolerance with periodic ammonia intoxication." Lancet, 1:1014-15, 1964.

Colombo, J.P; W., Burgi; R. Richterich et al.: "Congenital lysine intolerance with periodic ammonia intoxication: a defect in L- -lysine degradation." Metabolism, 16:910-25, 1967.

JAKSCH-HAYEM SYNDROME

Synonymy

S. of pseudoleukemic anemia. S. of Jaksch-Hayem-Luzet. S. of infantile splenic anemia.

Syndromography

Clínic

It occurs in children under 3 years of age. It is characterized by weakness, gastrointestinal disorders, irregular fever.

Physical examination. Paleness, hepatosplenomegaly, and lymphadenopathy are noted. Growth retardation.

Paraclinical examinations

Blood. Anemia, anisopolkylocytosis, pseudoleukemic leukocytosis with relative lymphocytosis. Normoblasts. Increase in the number of reticulocytes. Marked thrombopenia.

Syndromogenesis and etiology

It represents a complex syndrome that recognizes a wide variety of etiological agents from malnutrition to infections. Thalassemia, hemolytic diseases of the newborn.

Literature

Von Jaksch, R.: Uber Leukamie und Leukocytose im Kindesalter", Wien. Klin. Wschr., 2:435-437, 456-458, 1889.

JARCHO-LEVIN SYNDROME

Synonymy

Spondylocostal dysplasia. Spondylothoracic dysplasia. Costovertebral dysplasia. S. spondylothoracic.

Syndromography

Clinic

In these newborns, death occurs before one year due to respiratory infections, or in early childhood.

Physical examination. They have a very short neck and trunk in contrast to the limbs. Among the vertebral abnormalities are: scoliosis and kyphoscoliosis, winged scapulae. Limitation of neck movements.

Patients have a prominent occiput, broad forehead, wide nasal bridge, anteverted nostrils, and short thorax. They give the impression of long fingers with camptodactyly and syndactyly.

Other abnormalities have been reported such as: hydronephrosis with urethral obstruction, bilobed bladder, absence of external genitalia, analyurethral atresia, cerebral polygyria.

Paraclinical examinations

Radiology. The shortness of the thorax is confirmed with decreased ribs and multiple vertebral defects. Hemivertebrae at practically all levels of the spine. Fused costal arches.

The rest of the skeleton is normal.

Ultrasound. In search of the described malformations.

Syndromogenesis and etiology

There is a wide variability in affected members of a family; in some cases spina bifida and open meningocele have been reported.

Two subtypes are suggested: spondylocostal and spondylothoracic

The term dysostosis better defines the defect, since the bones not involved are normal.

Genetic etiology and autosomal recessive inheritance. The defect The basics are unknown.

Literature

Karnes, P.S. et al. : Jarcho-Levin's syndrome: four new cases and classification of Subtypes". Am. J. Med. Genet., 40:264-270, 1991.

Poor, M.A. et al.: "Nonskeletal malformations in one of three siblings with Jarcho-Levin syndrome of vertebral anomalies."
J. Pediatr., 103:270-272, 1983.

JOHANSON-BLIZZARD SYNDROME

Synonymy

S. of aplasia of nasal wings.

Syndromography

Clinic

In those affected by this syndrome we find poor growth from the prenatal stage. Mental development ranges from severe mental retardation to normal intelligence. From the neurological point of view there is sensory deafness and hypotonia.

A craniofacial dysmorphic pattern characterized by mild to moderate microcephaly is described. Posterior midline scalp defects. Hypoplasia or aplasia of the nasal wings. Sparse and scattered hair in the frontal region.
Lacrimonasal duct fistulas. Lack of some permanent teeth and hypoplasia of the temporary ones.

Occasionally we can observe strabismus and aplasia of the lacrimal punctum.

In the urorectal region they present imperforate anus, double or septate vagina. Cryptorchidism. Single urogenital orifice. Micropenis. Caliectasias to hydronephrosis. Recto-vaginal fistula.

In the endocrine sphere, primary hypothyroidism is reported in a third of cases and an evolution by degrees. On the other hand, there is pancreatic insufficiency with intestinal malabsorption and anemia.

Among other occasional anomalies we have small nipples and absence of the areola, radiolucent defects of the skull and abdominal and thoracic situs inversus.

Paraclinical examinations

Blood. Hemogram. Iron deficiency and low hemoglobin.

Auditory evoked potentials.

GDP. Diminished. Low total proteins. Serum calcium: hypocalcemia.

Cholesterolemia. Normal

Electroencephalogram. Abnormal.

Enzymatic studies. Absence of trypsin, chymotrypsin, amylase, carboxypeptidase and lipase.

Syndromogenesis and etiology

Of genetic etiology with autosomal recessive inheritance and unestablished basic defect. Pathological studies have shown pancreatic changes in which fat replacement with insufficient acinis is observed without concomitant hypoglycemia and in the brain cortical neuronal disorganization and abnormal gyral formation.

Literature

Day D.W. and J.N. Israel: "Johanson-Blizzard's syndrome" Birth Defects, 14 (6B): 275-287, 1978.
Moeschler, J.B. and M.S. Lubinsky: “Brief clinical report:Johanson- -Blizzard's syndrome with normal intelligence." Amer. J.Med. Genet., 22:69-73, 1985.

JULIEN-MARIE-SEE SYNDROME

Synonymy

S. of hydrocephalus-hypervitaminosis.

Syndromography

Clínic

It appears in children 24 hours after administration of a single, massive dose of vitamins A and D (300,000 IU or more of vitamin A).

It is characterized by the presence of vomiting and drowsiness. Diplopia, papillary edema and other symptoms that raise suspicion of a brain tumor (pseudotumor cerebri) may also occur.

Physical examination. There is sudden and intense bulging of the fontanela and the rest of the examination is negative.

Paraclinical examinations

Lumbar puncture. It's normal. Within 24 hours all signs and symptoms disappear spontaneously.

Syndromogenesis and etiology

Acute vitamin A toxicity.

It is the equivalent of the headache reported in adults receiving massive doses of vitamin A.

Literature

Marie J. and G. See: "Hydrocéphalie aigue benigne du nourrisson apres ingestion d'une dose massive et unique de vitamines A et D." Arch. Franc. Pédiat., 8:563-65, 1951.

KLOEPFER SYNDROME

Syndromography

Clinic

It appears in both sexes and begins around 2 months of age. It is characterized by outbreaks of blisters or severe blisters after sun exposure. It is accompanied by complete blindness, imbecility with progressive dementia. There is growth arrest between 5-6 years of age coinciding with a decrease in erythema. Patients generally die in the second decade of life.

Syndromogenesis and etiology

Autosomal recessive inheritance.

The etiology has not been specified.

Literature

Kloepfer, J.W. Quoted by V.A. McKusick: Medical genetics, 1958-1960. St. Louis, Mosby, 1961, p. 24.

KOSTMANN SYNDROME

Synonymy

Genetic infantile agranulocytosis S. S. of congenital neutropenia. Childhood lethal agranulocytosis S.

Syndromography

Clínica

This familial disease is characterized by the onset, in the first months of life, of pyogenic, recurrent and severe infections, especially in the skin and lung.

Signs on physical examination depend on the location of the infection. There is no splenomegaly.

The prognosis is unfavorable, death generally occurs due to severe infections such as meningitis.

Paraclinical examinations

Blood. Number of normal leukocytes with absolute or very marked neutropenia, absolute and relative eosinophilia and monocytosis. There is no anemia or it is only secondary in the late periods of the disease. The number of platelets is normal.

Bone marrow. Normal erythropoiesis. Normal megakaryocytes. Myeloid series: absence of myeloid precursors from the early stages. Epinephrine stimulation test, Window test (Rebuck): reveals absence of neutrophils.

Pathological anatomy. Necrotic ulcerations in the genital mucosa. Diffuse inflammatory reaction with leukocytes, plasmacytes and histiocytes, without neutrophils in several tissues. Liver and spleen moderately congested. Signs of extramedullary hematopoiesis in various tissues.

Syndromogenesis and etiology

The basic enzymatic or metabolic defect is unknown.

Autosomal recessive inheritance has been suggested.

Literature

Kostmann, R.: "Infantile genetic agranulocytosis (agranulocytosis infantile hereditaria): new recessive lethal disease in man." Paediat Act. (suppl. 105), 45:1-78, 1956.

KUNDRAT SYNDROME

Synonymy

S. of arrhinoencephaly. S. of holoprocencephaly. (See trisomy D1 syndrome).

Syndromography

Clinic

Same as Dl trisomy syndrome, except for the absence of the typical dermatoglyphic pattern.

The prognosis is poor, they die in early childhood.

Paraclinical examinations

Genetic study. There are generally 46 chromosomes. Short arm of chromosome 18 or other chromosomal abnormality should be explored.

Syndromogenesis and etiology

Several genetic mechanisms have been considered.

The etiology is unknown.

Literature

Kundrat, H.: “Archinencephalie als typische Art von Missbildung.” Vienna. Med. Bl., 5:1395, 1882.

LACRIMO-AURICLE-DENTO-DIGITAL SYNDROME

Synonymy

S. LADD. by Levy-Hollister.

Syndromography

Clinic

As its name indicates, this syndrome is characterized by lacrimal anomalies consisting of the absence or decrease in unilateral or bilateral tears or chronic lacrimation, chronic or recurrent conjunctivitis or dacryocystitis, fistulas of the nasolacrimal duct and absence or hypoplasia of lacrimal glands or lacrimal puncta, lacrimal sac or nasal passage.

Atrial defects consist of cup-shaped atria with or without sensorineural deafness in about 70% of reported cases.

The teeth are spike-shaped, there may be agenesis of salivary glands that is manifested by xerostomia and/or absence of the Stensen duct or papilla.

Digital anomalies consist of a bifid or hypoplastic triphalangeal thumb, syndactyly of the second, third and fourth fingers, hypoplasia of the hypothenar area, radio-ulnar synostosis, and radial aplasia. In addition, genitourinary anomalies have been reported.

Paraclinical examinations

Radiology. The referred bone defects are checked.

Kidney ultrasound. Diagnosis of kidney anomalies.

Hearing tests. Detection of hearing loss.

Syndromogenesis and etiology

This syndrome has a genetic etiology with autosomal dominant inheritance; it has been suggested that this syndrome represents an incomplete form of expression of the EEC syndrome.

Literature

Gorlin, RJ.: M.M. Cohen and L.S, Levin: Syndromes of head and neck. Oxford Monographs on Medical Genetics., No.19. Oxford, 1990 p. 868.

LANGER-GIEDION SYNDROME

Synonymy

S. of acrodysplasia with exostoses. S. of thoracic-pelvic-phalangeal dystrophy. S. tricho-rhino-phalangeal with exostoses.

Syndromography

Clinic

There is a dysmorphic pattern characterized by moderate microcephaly. Large ears laterally protruded. Thick eyebrows. Sunken eyes. Large, round nose with thick nasal wings and septa, angled nostrils and wide nasal bridge. Long and prominent upper lip and microretrognathia. Occasionally, ocular hypotelorism, eyelid ptosis, prominent eyes, coloboma of the iris and epicanthus are included. Sparse and fine hair. Divergent strabismus.

Paraclinical examinations

Radiology. Flattening of the condyles of the mandible.

Cone-shaped epiphysis. Narrow costal arches posteriorly, winged scapulae, growth retardation, multiple exostoses.

Karyotype. They may present deletion of chromosome 8: of (8)(q24.11 and q24.13).

Syndromogenesis and etiology

Although the majority of reported cases have been sporadic, autosomal dominant inheritance is proposed, since transmission from male to male has been seen. Both normal karyotypes and those with 8g deletion have been reported. The basic defect is unknown.

Literature

Hall, B.D. et al.: "Langer-Giedion syndrome." Birth Defects. 10(12): 147-164, 1974.

Okuno, T. er al.: "Langer-Giedion syndrome" wiht 8 (q24.13- q24.22). Clin. Genet., 32:40-45, 1987.

LARSEN-JOHANSSON SYNDROME

Synonymy

S. de Larsen. S. cleft palate-flat face-multiple dislocations.

Syndromography

Clinic

These patients have a flattened face with depression of the nasal bridge, bulging forehead, and hypertelorism. Cleft palate appears in 30% of cases, almost always limited to the soft palate or uvula; some patients may have laryngotracheomalacia. The most characteristic of the syndrome are dislocations of the elbow, hip, knees, and wrists, the fingers are long with a spatulated thumb, and the club feet are valgus or varus. The nails are small. With orthopedic treatment they are able to walk late and osteoarthritis of the large joints is a potential complication.

Paraclinical examinations

Radiology. Dysplastic development of epiphyseal centers, multiple body ossification centers. Delayed coalescence of the calcaneal ossification centers.

Syndromogenesis and etiology

The etiology is not well defined; the autosomal dominant monogenic etiology with very variable expressivity has been assessed.

Literature

Larsen, L.J.; E.R. Schottstaedt and F.C. Bost: “Multiple congenital dislocations associated with characteristic facial abnormality.” J. Pediatr.. 37:674-581, 1950.

Stanley, D. and N. Seymoor: The Larsen's syndrome occurring in four generations of one family." Int. Orthop., 8:267-272, 1985.

LEIGH SYNDROME

Synonymy

Childhood subacute necrotizing encephalopathy. S. de Wernicke in childhood.

Syndromography

Clinic

It occurs equally in both sexes, beginning in early childhood during the first year of life. It is characterized by slow development, with slight hypotonia, loss of head control, poor sucking, anorexia and vomiting, irritability and continuous crying, generalized seizures and myoclonic contractions. If it begins during the second year of life, it is manifested by ataxia, dysarthria, intellectual involution, tonic spasms, characteristic respiratory disorders (episodes of hyperventilation, periods of apnea, sighing, sobbing without crying), external ophthalmoplegia and abnormal eye movements (irregular rotation, peduncular nystagmus), paralysis of swallowing, abnormal movements of the limbs of type ataxic and choreiform.

Paraclinical examinations

Urine.

Blood.

Cerebrovascular fluid.

Syndromogenesis and etiology

It is a syndrome of genetic etiology and is transmitted by autosomal recessive inheritance. It is caused by a deficiency of the enzyme pyruvate carboxylase (PC). PC is an allosteric enzyme of four identical subunits, almost totally dependent on acetyl COA as an activator. It is very abundant in astrocytes but is low or absent in neurons.

It is known that neurotransmitters accumulate in presynaptic terminals, partly due to the de novo synthesis that takes place at this site and also because glutamine, metabolically derived from astrocytes, is the major precursor of glutamate and GABA (gamma-aminobutyric acid). In turn, the formation of glutamine occurs using the carboxylation of pyruvate that takes place in astrocytes as an initial step.

The loss of PC activity determines a decrease in glutamine, as well as depletion of intermediate carbons 4 and 5, which determines the death of the neuron, in addition to affecting the synthesis of myelin, including the absence of its formation. The defect in these two actions of PC in brain metabolism may explain the pathology of the central nervous system in this syndrome.

There appears to be genetic heterogeneity in the CP defect, since three clinical forms are described:

1. Simple form, which presents in the first month of life, with moderate lactic acidemia and moderate psychomotor retardation.
2. Complex form, which appears after birth with complex lactic acidemia, accompanied by hyperammonemia, citrullinemia and hyperinsulinemia, which causes death before three months of age.
3. Semi-simple form, which only presents episodes of acidosis without mental retardation.

Literature

Leigh, D.: "Subacute necrotizing encephalomyelopathy in an infant." J. Neurol. Neurosurg. Psychiat., 14:216-21, 1951.

Scriver, C.R. et al.: The metabolic and molecular bases of inherited disease. 7th edition. McGraw-Hill, Inc. New York, 1995.

LENZ SYNDROME

Synonymy

S. of microphthalmia-polyformations.

Syndromography

Clínica

It is characterized by: intense, bilateral microphthalmia with congenital blindness. Skeleton: sloping shoulders, narrow due to clavicular defects; thorax cylindrical, long; lumbar lordosis; double thumbs and stubby fingers. Teeth: agenesis of some lateral incisor. Genitourinary: hypospadias, cryptorchidism. Other occasional anomalies: congenital heart disease, mental retardation, physical retardation, small stature.

The ophthalmological manifestations are: colobomatous microphthalmos, blepharoptosis, nystagmus, strabismus.

Paraclinical examinations

Radiology. Renal agenesis, hydroureters.

Congenital heart disease. Skeletal alterations.

Syndromogenesis and etiology

It only occurs in males, due to inheritance linked to the X chromosome, with slight expressiveness in female carriers of the mutation.

The etiology is genetic.

Literature

Goldberg, M.F. and V.A. McKusick: X-linked colobomatous microphthalmus and other congenital anomalies". Am. J. Ophthalm., 71:1128, 1971.

Herrmann, J and J.M. Opitz: The Lenz microphthalmia syndrome. Birth Defects. New York, Nat. Found.. 1972, p.138.

Lenz, W.: "Recessiv-geschlechtsgebundene Mikrophtalmie mit multiple Missbildungen." Z. Kinderheik., 77:384, 1955.

LEROY SYNDROME

Synonymy

S. of I-Cell. S. of mucolipidosis type I.

Syndromography

Clinic

Clinical manifestations have an early onset. The birth weight is less than 5.5 lb and there is marked growth deficiency.

Patients have a characteristic facial dysmorphism in which a high and narrow forehead, thick eyebrows, bloated eyelids, epicanthus, depressed nasal bridge, anteverted nostrils, long philtrum, and progressive alveolar hypertrophy stand out. They present limitations in dorsolumbar movement, increased volume of the wrists and fingers. The skin is thick and relatively tight during childhood. Cavernous hemangiomas are common. At 18 months they are able to sit with support and some also stand with help, however, the delay in growth and development is severe and progressive. Respiratory infections (bronchitis, pneumonia) and otitis media occur with high frequency.

Death occurs at 5 years associated with heart failure.

Paraclinical examinations

Radiology (bone survey) in which severe bone alterations similar to those of Hurler syndrome stand out.

Peripheral lymphocytes and fibroblast culture. They contain abundant cytoplasmic inclusions.

Acid hydrolase activity. Considerably increased

mentioned.

Syndromogenesis and etiology

Of autosomal recessive monogenic etiology.

There is a notable increase in lysosomal enzymes in the plasma.

In the cerebrospinal fluid and urine, lysosomal deficiency is found in cultures of fibroblasts and of the enzyme N- -acetylglucosamine-1-phosphotransferase, this apparently being the basic defect of the syndrome.

Literature

Wiesmann, U. et al.: "Mucolipidosis I ((-cell disease). A clinical and biochemical study." Acta Paediatr. Scand., 63:9-16, 1974.

LETTERER-SIWE SYNDROME

Synonymy

S. by ABT-Letterer. S. of Siwe.

Syndromography

Clinic

It occurs in growing children (below 3 years of age), occasionally later in young adults. It is characterized by asthenia, anorexia, irritability, exhaustion, chronic otitis media, and persistent low-grade fever.

Physical examination. On the scalp, face and trunk: maculopapular skin lesions, with bright yellow or brown papules, with a reddish border and yellow center. Erosions with secretions from the skin folds, ecchymoses and petechiae. Edema, generalized lymphadenopathy and hepatosplenomegaly.

Paraclinical examinations

Blood. Leukocytosis, anemia, thrombocytopenia.

Radiology. Destructive bone lesions, especially in the thorax.

Syndromogenesis and etiology

Seven cases have been reported in family members. It is associated with histiocytosis X syndrome (see).

The etiology is unknown.

Bibliography

Letterer. E.: “Aleukamische Retikulose”. Frankfurt. Ztschr. F. Path.. 30:377-394, 1924.

Petersen, R.A. and T. Kuwabara: "Ocular manifestations of familial lymphohistiocytosis." Arch. Ophthal., 79:413-416, 1968.

Shoeck, V.W., R.D.A. Peterson, and R.A. Good: "Familial occurrence of Letterer-Siwe disease." Pediatrics, 32:1055-1063, 1963.

LAZY LEUKOCYTE SYNDROME

Synonymy

Lazy-leukocyte S. S. of inhibition of leukotaxis in recurrent infections.

Syndromography

Clínic

This syndrome was described by Miller, Oski and Harris in 1971. It begins around 6 months with recurrent infections:

stomatitis, gingivitis, otitis media, pyodermias and bronchopneu-

monias

Paraclinical examinations

Blood. Leukopenia and neutropenia (5%). Test for epinephrine, cortisone and endotoxin, negative. Negative chemotaxis (chemotaxis) in vertical microhematocrit tubes.

Rebuck window test. It shows absence of inflammatory exudate on the slide applied to the scarified skin, which expresses alteration of leukocyte mobility.

Syndromogenesis and etiology

Leukocytes react with difficulty to chemotactic stimuli. Patients with this syndrome have normal neutrophils in the bone marrow, but have abnormal random migration and abnormal chemotaxis.

The defect appears to be in the capacity for locomotion from the bone marrow to the circulation.

It is transmitted by autosomal recessive inheritance.

The etiology is genetic.

Literature

Miller, M.E.; FA. Oski and M.B. Harris: Lazy leukocyte syndrome: A new disorder, of neutrophil function." Lancet, 1:665, 1971.
Constantinopoulos, A.P. Nicolaidou; P. Mounis et al.: "Lazy leukocyte syndrome." J. Pediat., 87:945, 1975.

LIGHTWOOD SYNDROME

Synonymy

S. of renal tubular acidosis. S. of congenital hyperchloremic acidosis. S. of nephrocalcinosis. S. of Albright I. S. of Butler.

Syndromography

Clinic

This syndrome begins in childhood with anorexia, vomiting, constipation, apathy, irritability, polydipsia and polyuria. It persists in adults and is frequently associated with osteopathies, with osteoscopic and joint pain, gait alterations and eventually deformities.

Physical examination. There is growth retardation in the child, dehydration crisis, hypotonia and high blood pressure.

Paraclinical examinations

Blood. Hyperchloremia, hypokalemia, hypophosphatemia, slightly elevated urea, decreased pH. Normal or low calcium and sodium levels. Decreased plasma bicarbonate. Frequently hyperglobulinemia.

Urine. Alkaline, hypercalciuria, decreased or absent ammonia, hyperphosphaturia, leukocyte casts.

Descending pyelography. Fine calcifications (nephrocalcinosis) in the kidney and nephrolithiasis.

Syndromogenesis and etiology

The etiology is variable. More than 72 familial cases have been reported, and two types have been differentiated: one seems to correspond to autosomal dominant inheritance, of variable expressivity. The other is transmitted by autosomal recessive inheritance.

This syndrome is associated with various conditions such as: Besnier-Boeck-Schaumann, Epstein, Kahler-Bozzolo, Toni Fanconi, Sjögren, Lowe syndromes, as well as uretero-sigmoid transplantation, idiopathic hyperglobulinemia and myopathic syndromes.

Whatever the type or etiology, the essential pathogenic mechanism lies in the alteration of the factors that intervene in the reabsorption of bicarbonate and the acidification of urine and is fundamentally caused by the deficiency of carbonic anhydrase. Other criteria maintain that deficient ammoniagenesis with greater elimination of cations (sodium, potassium and calcium) would be the essential mechanism. Both hypotheses are considered that could explain the two main varieties and their frequent association.

Literature

Bennett, W.M. K.H. Hempel; J.E. Berland et al.: "Renal tubular acidosis." Arch Int Med. 121:81-86. 1968.
Lightwood, R.: *Calcific infarction of the kidneys in infants" Communication, Proc. Brit. Paediat. Soc. Dis. Child., 10:205- -206. 1935.

Nash, M.A.; AD Roasted; I. Greifer et al.: "Renal tubular acidosis in infants and children." J. Pediatr., 80:738, 1972.

CONGENITAL PULMONARY CYSTIC LYMPHANGIECTASIA

Syndromography

Clínica

It occurs in newborns (some stillborn) sometimes on the first day of life. There is cyanosis and respiratory distress.

Physical examination. Numerous rales in both lungs and sometimes associated pleural effusion. Lymphoedema of other organs can frequently be associated. Sometimes it is associated with cretinism or congenital cardiovascular diseases.

Paraclinical examinations

Chest radiology. Diffuse opacity in both lung fields. Associated pleural effusion.

Syndromogenesis and etiology

Primary congenital developmental disorder.

The etiology is unknown.

Literature

Fronstin, M.H.; G.S. Hooper: B.E. Besse et al.: "Congenital pulmonary cystic lymphangiectasis." Amer. J. Dis. Child. 114:330-35, 1967.
Virkow, R.: Gesammelte Abhandlungen zur Wissenschaflichen Medicin. Frankfurt, 1851.

LISENCEPHALY SYNDROME

Synonymy

S. of lissencephaly type I. S. of Miller-Dieker.

Syndromography

Clinic

In this syndrome there is a prenatal history of polyhydramnios in 30% of cases, decreased fetal movements, and newborns are small for gestational age.

In the perinatal period they present hypotonia, but 6 weeks later they are hypertonic children with evident opisthotonus. TO

At 9 weeks they develop seizures and death occurs around 6 months in half of the patients. Craniofacial dysmorphism is recognized by microcephaly with prominent occiput, large anterior fontanelle, and bitemporal depression. The skin in the glabella region is wrinkled.

The upper lip is thin, the nose is small and with the nostrils in anteversion, the ears are low set with posterior rotation and abnormal helix. Congenital heart disease has been reported in 65% of cases. Urogenital malformation, corneal opacity, polydactyly and camptodactyly. Lissencephaly has been described in the autopsies of these cases.

Paraclinical examinations

Ultrasound, computed axial tomography or magnetic resonance imaging. Dilation of the ventricles with thickened cortex and decreased white matter, thin or absent corpus callosum, moderate calcifications.

Karyotype0. 17p 13.3 deletion in more than 50% of cases.

Syndromogenesis and etiology

Lissencephaly may occur alone as a malformation sequence, as part of a complex malformation syndrome, or may be associated with other structural defects of the brain and craniofacial region not explainable by a migration defect. Lissencephaly has been defined as a complete degree (grade 1), almost complete (grade 2), or as a mixed agyria/pachygyria (grade 3). Its etiology has been referred to as a microdeletion syndrome, small deletion syndrome, and continuous gene syndrome.

Literature

Dobyns, W.B.: "Developmental aspects of lissencephaly and the lissencephaly syndrome." Birth Defects, 23 (1): 225-241, 1987.

ASYMMETRIC CRYING SYNDROME

Synonymy

Cardiofacial S.

Syndromography

Clínic

This syndrome is characterized by asymmetric crying facies, congenital heart disease, genitourinary and respiratory system malformations. Its frequency is 1/120 to 1/160 of newborns. Asymmetry is more common on the left side.

The most common heart diseases are ventricular septal defect, and tetralogy of Fallot, double aortic arch, pulmonary stenosis, coarctation of the aorta, atrial septal defect, common atrioventricular defect, tricuspid atresia, simple ventricle, hypoplastic right ventricle, hypoplastic pulmonary arteries and bicuspid aortic valve.

Vertebral anomalies, tracheoesophageal atresia or fistula, laryngomalacia and other respiratory system defects have also been reported.

Genitourinary malformations include renal agenesis or hypoplasia, ectopic kidney, polycystic kidney, and hypogonadism among the most frequent.

10% of cases have cleft lip and palate or isolated cleft palate.

Paraclinical examinations

Abdominal ultrasound. To look for the kidney malformations described.

Chest and cervical spine radiology. Vertebral anomalies include hemivertebra, fused vertebrae, sternum and costal anomalies.

Echocardiogram. For the diagnosis and characterization of the described heart diseases.

Syndromogenesis and etiology

Monogenic genetic etiology has been suggested and the spectrum of anomalies has been considered as an overlap of the oculoauriculovertebral spectrum with minimal expression.

Literature

Borlin, R.J.; M.M. Cohen and L.S. Levin: Syndromes of Head and Neck. Oxford Monographs on Medical Genetics No.19 Oxford, 1990, p. 613.

LOWE SYNDROME

Synonymy

Oculo-cerebro-renal syndrome.

Syndromography

Clínica

This syndrome is characterized by hypotonia, cataracts, and renal tubular dysfunction.

Hypotonia is accompanied by joint hypermobility that progresses to joint contractures; cataracts with or without glaucoma and renal tubular dysfunction, with limited ammonia production and hyperchloremic acidosis, phosphaturia, tendency to hypophosphatemia and generalized aminoaciduria, as well as albuminuria and organic aciduria.

Signs of rickets Increase in joint volume, synovial thickening.

Occasionally there may be convulsions, cranio-synostosis.

Paraclinical examinations

Radiology. Bone signs of rickets, osteoporosis.

Urine. Metabolic tests (aminoaciduria): phosphorus (phosphaturia).

Studies of renal tubular function. Renal acidosis.

Electroencephalogram. Abnormal.

Syndromogenesis and etiology

Genetic etiology and autosomal recessive inheritance linked to the X chromosome. Although there is renal acidosis, there is evidence that the basic defect may involve an alteration of the connective tissue.

Literature

Tripathi, R. et al.: “Lowe's syndrome". Birth Defects Orig. Art. Ser.. 18:629-644. 1982.

LUDER-SHELDON SYNDROME

Synonymy

S. of familial defect of tubular absorption of glucose and amino acids.

Syndromography

Clínica

It affects both sexes from childhood. Growth retardation

I lie.

Osteomalacia or rickets may be seen.

Paraclinical examinations

Urine. Excessive excretion of several amino acids. Glucose, fructose and proteins are also excreted.

Syndromogenesis and etiology

Defect in tubular function with excess flow of amino acids and glucose but not phosphates. Autosomal dominant inheritance.

Literature

Luder, J. and Sheldon, W.: Familial tubular absorption defect of glucose and amino acids." Arch. Dis. Child., 30:160-64, 1955.

Gallien, R.: "Maladies a erreur métabolique et hyperaminoaciduries." Laval Med., 37:48-68, 1966.

MALABSORPTION SYNDROME OF METHIONINE

Synonymy

S. of oast-house (hop dryer).

Syndromography

Clinic

It begins in the infant with a picture of chronic diarrhea with abundant stools, which have excessive weight and increased nitrogen. Urine: sweet hop smell, produced by its high alpha-hydroxybutyric acid content. Central nervous system: convulsions in the form of extension spasms;
mental retardation and febrile seizures with tachypnea and edema. Appearance: peculiar with dry skin, blue eyes and white hair.

They improve when methionine in the diet is reduced.

Paraclinical examinations

Urine. Increased alpha-hydroxybutyric acid. The administration of methionine triggers diarrhea.

Syndromogenesis and etiology

It is due to a failure in the transport of methionine through the intestinal epithelium: intestinal bacteria transform it into various metabolites, including alpha-hydroxybutyric acids.
alpha-ketobutyric and alpha-aminobutyric acid that pass into the blood and are eliminated through urine, giving it the sui generis odor, some other metabolite damages the nervous system, causing seizures and mental retardation.

It is transmitted in an autosomal recessive manner.

The etiology is genetic.

Literature

Frederickson, J.B. In: Stanbury-Wyngaarden-Frederickson: The Metabolic Basis of Inherited Diseases. ed. 3, Ed. McGraw-Hill, New York, 1972. p. 1500.

Hoof, C.; J. Timmermans; J. Snoeck et al. : "Methionine malabsorption n in a mentally child." Lancet, 2:20, 1964.

Hooft, C.; J. Timmermans; J. Snoeck et al.: "Methionine malabsorption syndrome." Ann. Paediat., 205:73, 1965.

Smith, A.J. and LB. Strang: “An inborn error of metabolism with the urinary excretion of alfa-hydroxybutyric acid and phenipyruvic acid." Arch. Dis. Child., 33:109, 1958.

MANNOSIDOSIS SYNDROME

Synonymy

Hurler-like storage system.

Syndromography

Clinic

It begins in the first year of life. They are tall and malnourished children with a tendency to respiratory infections.

Physical examination. Prominent forehead with coarse features, macroglossia, large ears; macrocephaly; moderate hepatomegaly, sometimes small splenomegaly, which later disappear; somatopsychic delay, muscular hypotonia, Babinsky, hyperreflexia, seizures, deafness: cataracts, clear coneas, blurred and gray optic papillae.

Paraclinical examinations

Blood. Vacuolated lymphocytes.

IgG. Diminished.

IgA. Absent.

Urine. Marked mannosuria.

Electroencephalogram. Normal.

Radiology. Thick skull cap, bone dysostosis.

Syndromogenesis and etiology

Genetics: autosomal recessive. Deficiency of alpha-mannoxidase in the liver, spleen and brain produces retention of mannose, which is deposited in the viscera.

Literature

Alusworth, A.S.; HA. Taylor; C.M. Stuart et al.: "Mannosidosis Phenotype of a severely affected child and characterization of alpha-mannosidase activity in cultured fibroblasts from the patient and his parents." J. Pediat., 88:814, 1976.

Ockerman, P.A.: A generalized storage disease resembling Hurler's syndrome." Lancet, 2:239, 1967.

Tsay, GL.; G Dawson and R. Matalon: *Excretion of mannose-rich complex carbohydrates by a patient with alfa-mannosidase deficiency ('mannosidosis')". J. Pediat., 84:865, 1974.

MARCHESANI SYNDROME

Synonymy

S. of congenital dysmorphodystrophy mesodermalis. S. de Weil- - Marchesani. Inverted Marfan syndrome with spherophakia and brachymorphia. S. of brachydactyly-spherophakia.

Syndromography

Clinic

They are short patients, with a wide skull, narrow orbits, slight maxillary hypoplasia with a narrow palate. Ocular symptoms begin in the first decade, with myopia, with or without glaucoma, ectopia lentis in 50% of cases and blindness in a third of cases.

Malformed teeth. Hands with brachydactyly. It is impossible for the patient to close the fingers of his hands completely and turn them into a

Congenital heart disease has occasionally been reported. Intelligence is not affected.

Physical examination. Eyes: spherophakia, microphakia, lens subluxation. Short stature, average 148 cm, short limbs, brachydactyly. In some cases heart murmurs.

Paraclinical examinations

Radiology. Hands, symmetrical shortening with open metacarpals and phalanges and delayed carpal ossification. The feet, including the toes, can participate in the same process.

Slit lamp eye examination. Good medicated mydriasis.

Syndromogenesis and etiology

It is considered a basic defect of embryological mesodermal development. Marchesani considers this syndrome to be the opposite of Marfan's and represents the late hyperplastic variant (brachydactyly).

The etiology is monogenic genetics with autosomal recessive inheritance. Genetic heterogeneity or the possibility of pseudodominance due to deletion of the dominant allele and expression of the recessive mutation on the homologous chromosome has been suggested.

Literature

Feiler-Ofry, V; R. Stein and V. Godel: "Marchesani's syndrome and chamber angle anomalies." Amer. J. Ophthal., 65:862-866, 1968.

Gorlin, R.J.; PR. L'Henreux and I. Shapiro: "Weill-Marchesani's syndrome in two generations: Genetic heterogeneity or pseudodominance." J. Pediatr. Ophthalmol., 11:139-145, 1974.

Marchesani, O.: "Brachydaktylie und angeborene Kugellinse als Systemar-Krankung." Kein. Mbl. Augenh., 103-392-406, 1939.

MARINESCO-SJÖGREN SYNDROME

Synonymy

S. of cerebellar ataxia-hypotonia-cataracts.

Syndromography

Clínic

Mild or moderate growth deficiency. Microcephaly, moderate to severe mental deficiency, cerebellar ataxia, weakness with or without hypotonia and tendency to nystagmus and dysarthria and cataracts evident from very early stages of life.

Paraclinical examinations

Eye examination. They verify the alterations described

Syndromogenesis and etiology

The etiology is genetic of autosomal recessive inheritance with a high incidence of consanguinity.

Pathological studies indicate a degenerative process in the cortical area of the cerebellum. Electron microscopy studies have revealed abnormally enlarged lysosomes containing spiral sheets or amorphous inclusion bodies.

Literature

Walker, P.D.; M.G. Blitzer and E. Shapira: "Marinesco-Sjögren's syndrome: Evidence for a lysosomal storage disorder." Neurology, 35: 415, 1985.

MAROTEAUX-LAMY SYNDROME

Synonymy

S. of mucopolysaccharidosis VI. S. of polydystrophic dwarfism.

Syndromography

Clinic

It affects both sexes, begins in childhood. The same clinical symptoms as in Hurler syndrome except intelligence, which is normal.

The onset of symptoms occurs between the first and third years of age when growth deficiency is evident, the facies becomes coarse with a large nose and thick lips, and the nasal bridge remains depressed. There is fine corneal opacity. The joints are prominent and limit movement. The skin is thick and umbilical and inguinal hernias occur. The teeth are separated and eruption is late. There is hepatos- plenomegaly and deafness, macroglossia, macrocephaly and over time the heart valves become involved.

Respiratory infections and diarrhea are frequent complications.

Paraclinical examinations

Radiology (bone survey). Wide and irregular metaphyses, these characteristics are also evident in the epiphyses. The vertebrae are flattened with anterior impingement of T-12 and L-1; The ribs are wide, the sternum prominent. Elongated Turkish chair. Odontoid hypoplasia, lumbar kyphosis and genu valgum.

Urine metabolic tests. Toluidine blue positive, mucopolysaccharide chromatography in 24-h urine, dermatan sulfate excretion.

Enzymatic studies. N-acetylgalactosamine-4-sulfatase (arylsulfatase B) deficiency.

Urine. Excessive excretion of chondroitinsulfate B.

Syndromogenesis and etiology

The basic defect consists of deficiency of the lysosomal enzyme arylsulfatase B, which determines intracellular accumulation of mucopolysaccharides that progressively affects the phenotype. Currently, two clinical forms are described according to the intensity of their manifestations: one severe and the other light.

Clinical defects are the consequence of the storage of mucopolysaccharides.

The etiology is genetic and the inheritance is autosomal recessive.

MARSHALL SYNDROME

Synonymy

S. of cataract-deafness-small nose with flat nasal bridge.

Syndromography

Clinic

Patients have short stature, a short and depressed nose with a flat nasal bridge and anteverted nostrils. Large looking eyes. Flat facial middle third. Upper incisors that protrude and are prominent. Thick lips. Mine-

pia, cataracts, esotropia. Sensorineural deafness. There may be mental retardation, retinal detachment and cleft palate.

Paraclinical examinations

Radiology (seo survey). Thick skull with absence of frontal sinuses, Intracranial calcifications. Spondyloepiphysial abnormalities with slight platyspondyly, irregular proximal epiphysis of the tibia and distal femur, bowing of the radius and ulna, wide ridges of the distal phalanges.

PEATC. Detection of deafness.

Syndromogenesis and etiology

Genetic etiology of autosomal dominant inheritance. Unknown basic defect.

Literature

O'Donnel, J.J.; S. Sirkin and B.D. Hall: "Generalized bone abnormalities in the Marshall's syndrome." Birth Defects Original Article Series, XII (5) : 299, 1976.

MARSHALL-SMITH SYNDROME

Synonymy

S. of accelerated skeletal maturation.

Syndromography

Clinic

These are newborns with a length greater than 50 cm who begin to have accelerated linear growth with accelerated skeletal maturation but with a weight lower than that which should correspond to their height. They present delayed psychomotor development and hypotonia and in the end they have an IQ. of 50. They present a dysmorphism characterized by a prominent forehead, narrow orbits with prominent eyes and blue sclerae, a nose with anteverted nostrils, a depressed nasal bridge, and small mandibular branches.

The hands have thick fingers on the first and second phalanges, the third being tapered. Hypertrichosis and umbilical hernia, choanal atresia, stenosis, or both are also observed; abnormalities of the larynx and laryngomalacia. Immune deficiency.

Most die from respiratory complications around 20 months.

Paraclinical examinations

Radiology. Accelerated bone age, at birth the ossification nuclei suggest an age of 4 years or more. The long bones tend to be thin but the proximal and middle phalanges are wide and the distal phalanges are thin.

Brain ultrasound or computed axial tomography. Abnormalities such as cerebral atrophy, macrogyria, agenesis of the corpus callosum.

Syndromogenesis and etiology

The reported cases have been sporadic in families and their etiology and basic defect are unknown.

Literature

Hoyme, H.E. and M.J. Bull: "The Marshall-Smith's syndrome: "Natural history beyond childhood". Clin. Research, 35:225A, 1987.

MCARDLE SYNDROME

Synonymy

McArdle-Schmid-Pearson type V.S. glycogen storage disease. S. of glycogenosis due to myophosphorylase deficiency. S. glycogenosis type Cori V.

Syndromography

Clinic

It usually begins in childhood although the diagnosis can be made later. First pain and then stiffness, after an exercise of any muscle, including the masseter muscles;
These alterations disappear with rest. In some cases, transient myoglobinuria may be observed.

In children they are a cause of hypotonia in the group of myopathies.

It has a fairly benign course.

Physical examination. Muscle size, tone and strength are normal when exercise begins. There are no fasciculations, fibrillation or myoclonus, normal reflexes. After three decades, atrophy and permanent weakness may appear in some muscles.

Paraclinical examinations

Electromyography. There may be increased activity.

Muscle biopsy. Glycogenosis.

Serial urine electrophoresis. Deletion of myglobins.

All routine laboratory tests are normal.

The venous return of the exercising muscle is poor in lactate and pyruvate, contrary to normal. The epinephrine test shows lower than normal lactate levels.

Syndromogenesis and etiology

It presents genetic etiology with autosomal recessive inheritance. It is a glycogen metabolic defect due to deficiency of the muscle glycogen-phosphorylase enzyme. Molecular studies have allowed the cloning of the muscle phosphorylase cDNA.

Literature

Gautron, S. et al.: "Molecular Mechanisms of MCArdle's disease (muscle glycogen phosphorylase deficiency)". J. Clin. Invest., 79:275-281, 1987.

McArdle, B.: "Myopathy due to a defect in muscle glycogen breakdown" Clin. Sc.. 10:12, 1951

Porte, D.; D.W. Crawford, D.B. Jennings et al.: *Cardiovascular and metabolic responses to exercise in a patient with McArdle's syndrome". New Eng. J. Med., 275:406-412,1966.

MCCUNE-ALBRIGHT SYNDROME

Synonymy

S. of osteitis fibrosa cystica.

Syndromography

Clínic

The patient presents bone deformities that progress during childhood, most commonly fractures of the upper end of the femur, the increase in bone density in the skull produces compressions that can cause blindness or deafness.

Precocious puberty that in women is characterized by menstruation before the development of breasts or pubic hair. There is acceleration of maturation coinciding with sexual precocity resulting in low stature. Irregular brown pigmentation, most commonly in the region of the sacrum, buttocks, upper spine and is unilateral in 50% of patients. In addition, there are other endocrine defects such as hyperthyroidism, hyperparathyroidism, Cushing's syndrome, acromegaly and hyperlactinemia.

Paraclinical examinations

Radiology. Multiple areas of fibrous dysplasia, most commonly in long bones and pelvis, in facial bones cause asymmetry. Bone deformities, with increased bone thickness.

Serum alkaline phosphatase. Elevated in 50% of cases.

Appropriate endocrine investigations.

Syndromogenesis and etiology

The reported cases are sporadic. Recent evidence suggests that endocrine abnormalities are the result of autonomic hyperfunction of several endocrine glands.

The etiology remains unknown.

Literature

Brunt P and V.A. McKusick: "Fibrous dysplasia. A report of genetic and clinical study with a review of the literature." Medicine, 49: 343-374, 1970.

D'Armiento, M. et al.: "McCune-Albright's syndrome: Evidence for autonomic multiendocrine hyperfunction." J. Pediatr., 102: 584-586, 1983.

MECKEL SYNDROME

Synonymy

S. de Gruber. S. de Meckel-Gruber. S. of splanchnocystic dysencephaly.

Syndromography

Clínic

Variable prenatal growth deficiency, posterior or dorsal encephalocele with mucocephaly and receding forehead, microphthalmia, cleft palate, micrognathia, ear anomalies, short neck, postaxial polydactyly, club foot, variable renal dysplasia with various degrees of cyst formation. Cryptorchidism, incomplete development of external and internal genitalia. Patients survive only a few days. Death occurs due to severe alterations of the nervous system and/or kidney defects.

Paraclinical examinations

Abdominal ultrasound. For observation of liver and kidney malformations and cysts, as well as malformations of the internal genitalia.

Brain ultrasound. Cerebral and cerebellar hypoplasia.

Karyotype. To exclude trisomy 13.

Syndromogenesis and etiology

Genetic etiology of autosomal recessive inheritance. Many of the clinical manifestations may be secondary to primary developmental defects.

Literature

Salonen, R.: "The Meckel's syndrome: Clinicopathological findings in 67 patients." Am. J. Med. Genet., 18:671-689, 1984.

MELNICK-NEEDLES SYNDROME

Synonymy

S. of osteodysplasty.

Syndromography

Clínica

On physical examination, this syndrome is characterized by the patient presenting abnormalities in gait and bulging of the upper limbs at the elbow; There may be a history of hip dislocation and delayed closure of the fontanels and they have a characteristic facies: the face is small, the eyes prominent, there is hypertelorism, a small jaw, and full cheeks. Although the size is usually normal, it is generally less than 10th. percentile. Patients tend to remain thin, with a small chest, pectus excavatum, narrow shoulders, and small breasts. The cases reported without diagnostic doubts have always been female. They cause dental malocclusion and over time osteoarthritis of the spine and hip are a complication.

Paraclinical examinations

Radiology. (bone survey). Increased density of the skull base, non-development of paranasal sinuses. Bowing of the radius and tibia. Distal widening of humeri, tibias and fibulas, coxa valga. Irregular costal arches, short clavicles.

The vertebrae are high with an anterior concavity in the thorax region. Flared iliac wings.

Syndromogenesis and etiology

The etiology is genetic due to a monogenic mutation but dominant inheritance linked to the X chromosome with lethality in males is assessed. The basic defect that explains this phenotype is unknown.

Literature

Barrio, A.L. M.C.M. Martínez: "Osteodysplasia. Melnick and Needles Syndrome", Radiology, 21:53-56. Madrid, 1979.

Gorlin, R.J. and J. Kmer: X-linked or autosomal dominant, lethal in the male, inheritance of the Melnick-Needles (osteo- dysplasty) syndrome?"

NEONATAL MIASTENLA GRAVIS SYNDROME

Synonymy

S. of transient neonatal myasthenia.

Syndromography

Clinic

It appears in newborns born to myasthenic mothers (10-20%). The disease in the mother can be very mild and not diagnosed early. The infant is weak and hypotonic

with poor suction, weak respiratory efforts, weakness in crying and swallowing, mask-like facies.

The prognosis is favorable with a good response to prastigmine and rapid recovery during the first weeks of life. In some cases there is persistence of symptoms; fatal cases have been reported.

Syndromogenesis and etiology

It is due to the passive transfer of anti-achr (Acetyl Choline Receptors, i.e. acetylcholine receptors) to the fetus from the myasthenic mother.

Literature

Stickroot, FL.; RL. Schaeffer and H.L, Bergo: "Myasthenia gravis occurring in an infant born of a myasthenic mother."
J.A.M.A., 120:1207-1209, 1942.

MONOSOMY 4P SYNDROME

Synonymy

S. de Wolf.

Syndromography

Clinic

Appreciable incidence. Sexual relationship IM:2F Dysmorphology.

Microcephaly. The nose has parallel edges and the root of the same width as the nasal tip.

The eyebrows are sparsely populated on the inner part, which gives a facial appearance of a 'Greek warrior helmet'. There is hypertelorism, the filter is narrow and deep with marked pillars that seem to protrude from the nasal passages. Lowered lip corners, small and retracted chin. Graceful limbs with dimples on elbows and knees. Long and pointed fingers. First thick and long toenail. Malformations. Cleft lip or palate, coloboma of the iris. Congenital heart disease in 50% of cases. Malformations of the central nervous system.

Neurology: Hypotrophy from birth. Severe mental retardation. Frequent seizures. Survival is little known.

Paraclinical examinations

Karyotype.
Electroencephalogram.
Computerized axial tomography.
Echocardiogram.

Syndromogenesis and etiology

Chromosomal imbalance due to deletion of the short arms of chromosome 4. In 10% of cases, there is a balanced translocation.

Literature

Leao, J.C. et al.: "New syndrome associated with partial deletion of short arms of chromosome No. 4". JAMA, 202:434-437, 1967.

Wolf, U. et al.: "Deletion on short arms of a B chromosome without cri du chat syndrome." Lancet, I:769,1965.

CONGENTTAL MORPHINISM SYNDROME

Synonymy

S. of narcotic deficiency in the neonate of an addicted mother.

Syndromography

Clínic

The alterations typical of this syndrome are evident between birth and the fourth day. They are newborns of low weight for gestational age, who present extreme irritability, with generalized tremors, excessive high-pitched crying, scratching and becoming hypertonic at the slightest tactile stimulus.

They can suffer seizures and lead to coma and death.

They do not suck, they cause vomiting and diarrhea, sneezing, respiratory distress, cyanosis, hyperthermia, lacrimation, seborrhea, sweating, insomnia, yawning and rarely jaundice.

This syndrome has a high mortality and consequences of brain damage.

Paraclinical examinations

Urine and blood. Determination of narcotics in mother and child.

Syndromogenesis and etiology

Maternal addiction to morphine, heroin, barbiturates and even methadone. The longer the period of addiction, the more frequent the syndrome is in the neonate, the closer to delivery the last dose, the earlier the syndrome appears. In addicted mothers it occurs with a frequency of 50-85%.

Literature

Kron, R.E.; M. Litt; M.D, Phoenis and colbs: "Neonatal narcotic abstinence effects of pharmacotherapeutic agents and maternal drug usage on nutritive sucking behavior." J. Pediat., 88:637, 1976.

Reddy, A.M.; R.G. Harper and G. Stern: “Observations on heroin and methadone withdrawal in the newborn". Pediatrics, 48:353, 1971.

Wasserman, E: In Gellis & Kagan: Current Pediatric Therapy-7:
Ed. Saunders, Philadelphia, 1976. p. 734.

MORQUIO SYNDROME

Synonymy

S. of atypical chondrodystrophy. S. of metaphyseal endochondral dysostosis. S. of mucopolysaccharidosis type IV.

Syndromography

Clinic

The onset is postnatal, between the first and third years of age, due to severe growth deficiency. Adults reach 82-115 cm.

They do not have mental retardation.

Physical examination. Eversion of the chondrocostal rim, genu valgum, kyphosis with anterior hump, carina chest; dwarfism with a height that does not exceed 120 cm, flat feet, short neck.

Facies: predominance of the lower facial third, no coarseness of features, small nose, anteverted and lateral nostrils.

thick bios. Minimal corneal opacity visible on slit lamp. Mixed conductive and sensorineural hearing loss.

They have a very short neck and their limbs have short hands, laxity of the wrist joint and small joints, genus valgus and deformity of the feet. Very short trunk with pectus carinatum and kyphoscoliosis.

Joint ligamentous laxity, restriction of mobility in the hips, normal intelligence. Dental enamel injuries.

The teeth are separated, with thin enamel and grayish color.

They have aortic regurgitation over time and hearing loss.

Inguinal hernia and hepatomegaly. Death occurs due to spinal cord compression or cardiac complications before the age of 20.

Paraclinical examinations

Urine. Keratosulfate excretion three or four times greater than normal.

Blood. Alder granules in leukocytes.

Radiology. In children under 2 years of age: oval vertebral bodies, hypoplasia of the odontoid processes of the axis, lumbar vertebrae with an anterior peak, long pelvis with wide iliac bones with anomalous acetabular and coxa valga ossification, metacarpals with a conical base. From 2-6 years: glossiform morphology of thoracic vertebrae and platyspondyly, compression of the femoral head. From 6-16 years: accentuation of radiological features and decrease in the number of carpal bones. Adults: progressive alterations of hips and mandibular condyles, more square vertebral bodies.

Urine metabolic tests. Toluidine blue is negative, but mucopolysaccharide chromatography shows keratin-sulfate excretion.

Study of enzymatic activity in skin fibroblasts or leukocytes. They show the enzyme deficiency.

Syndromogenesis and etiology

Of autosomal recessive genetic etiology, the basic defect is a deficiency of lysosomal enzymes. There are two types of Morquio syndrome: one that is clinically more severe and presents deficiency of N-acetylgalactose-mine-6-sulfatase (type IVA) and another that is clinically less severe with deficiency of the enzyme B- -galactosidase (type IVB).

These deficiencies affect the metabolism of mucopolysaccharides, which accumulate, causing the clinical picture described.

Congenital 6-sulfatase deficiency causes the formation of abnormal deposits in the chondroblasts, affecting bone growth, also in the coneas and occasionally in the myocardium (aortic insufficiency). Increased excretion of keratin sulfate through urine.

Literature

Blaw, M.E. and L.O. Langer: "Spinal cord compression in Morquio disease." J. Pediatr.. 74:593. 1969.

Langer, L.0. and C.S. Carey: "The roentgenographic features of Morquio disease." Amer. J. Roentgenol., 97:1, 1966.

Morquio, L.: "Sur une forme de dystrophie osseuse familiale." Bull. Soc. Pediat. Paris, 27:145, 1929.

MUCOVISCIDOSIS SYNDROME

Synonymy

Cystic fibrosis of the pancreas. Fibrocystic disease of the pancreas. (See cystic fibrosis CF.)

Syndromography

Clínica

Cystic fibrosis, described in the 1930s, is a multisystem condition characterized primarily by chronic airway obstruction and infection and malabsorption, with all its clinical consequences.

It begins in 10% of cases in the neonate as a meconium leak with absence of stool, abdominal bulge and mass in the right iliac fossa: the majority begins in the first months, with recurrent bronchopulmonary infections.

Respiratory system: coqueluchoid cough, emphysema, bronchopneumonic outbreaks and bronchial secretions with staphylococcus or E. Coli. Digestive: hyperorexia, foul-smelling voluminous stools, meteorism, often rectal prolapse. In older children, hepatosplenomegaly and portal hypertension. There is poor development and malnutrition with normal intelligence. Other occasional symptoms: thickening of the salivary glands; salty skin; clubbing in older children; hypoproteinemic edema.

Nasal polyposis, pansinusitis and hyperglycemia not related to diabetes mellitus.

Paraclinical examinations

Stool. Steatorrhea. Quantification of trypsin and chymotrypsin activity.

Sweat test. High sodium values (more than 60 mEq/L).

Radiology. Hypertransparent lung with irregular infiltrations.

Blood. Decreased serum pancreatic isoamylase.

Respiratory functional tests. Typical pattern of obstructive pulmonary involvement.

Syndromogenesis and etiology

The determining factors of the symptoms are: thick mucus and hyperconcentration of sodium chloride in sweat and saliva.

Dysfunction of the exocrine glands seems to be the most important pathogenic mechanism and is the cause of a set of broad manifestations. Pathological anatomy demonstrates characteristic changes in the mucus-secreting organs.

It is the most common lethal genetic disease in white children, occurring one case in every 1,600-2,000 live neonates. 15% of the population is an asymptomatic carrier.

It is transmitted in an autosomal recessive manner, both parents are heterozygous: it affects one in four children.

The etiology is genetic. The gene has been located on the long arm of chromosome 7, at position 7q31, which determines the production of the transmembrane conductance regulatory protein, responsible for alterations in the transport of chloride and sodium.

Literature

Andersen, D.H.: “Cystic fibrosis of the pancreas and its relationship to celiac disease.” Am. J. Dis. Child, 56:344, 1938.

Bramwell, A.: “A case of infantilism". Trans. Med. Chir. Soc. Edinburgh, 21:94, 1902.

Fanconi, G.: "Der intestinale infantilism und aehliche Formen des chronischen Verdaungsstoerung." Abh. Kiderheilk. Beiheft., 21:1, 1928.

HAPPY PUPPET SYNDROME

Synonymy

S. of children's dolls.

Syndromography

Clinic

Frequent attacks of infantile spasms; prolonged and easily provoked paroxysms of laughter; movements that imitate those of a doll or puppet; continuous protrusion of the tongue; mental deficiency.

Physical examination. Brachycephaly, microcephaly, occipital flattening or depression, prognathism, incomplete development of the choroid.

Paraclinical examinations

Skull x-ray. Vertical inclination of the base.

Electrocardiogram. Typical pattern expressed by: 1, absence of hypsarrhythmia. 2, presence of symmetrical and synchronous 2 cm/s wave and spike activity.

Syndromogenesis and etiology

Very rare form of childhood epilepsy.

The etiology is unknown.

Literature

Angelman, H.: "Puppet children, a report on three cases." Develop. Med. Child. Neurol., 7:681-88, 1965.

Bower, B.D. and P.M. Jeavons: "The 'happy puppet' syndrome." Arch. Dis. Child., 42:298-302, 1967.

Berg, J.M. et al.: "Angelman's 'happy puppet syndrome'." Am.J. Dis. Child., 123:72, 1972.

MULIBREY DWARF SYNDROME

Synonymy

S. of dwarfism with pericarditis. S. of Perheentupa.

Syndromography

Clínica

This syndrome is characterized by short stature, pericardial constriction, scattered yellow pigments in the fundus of the eye, hepatomegaly, hydrocephalus, the affected organs make up the term MULIBREY: MU-muscule; LI-liver (liver);
BR-brain (brain) and EY-eyes (eyes).

The growth defect has a prenatal onset, the hands and feet are large in relation to the body. Patients have a craniofacial dysmorphism characterized by a large dolichocephalic skull, high forehead, wide and depressed nasal root (hypertelorism), triangular facies, nevus flameus on the lip and chin.

They develop constrictive pericarditis with thick and adherent pericardium, hepatomegaly, ascites and dilation of cervical veins with venous hypertension. Intelligence is normal. Short survival due to pericardiopathy.

Paraclinical examinations

Radiology. Skull: J-shaped sella turcica, hypoplasia or aplasia of frontal and sphenoid sinuses. Fibrodysplasia mainly tibial; pericardial calcifications.

Ultrasound. Cerebral and abdominal.

Electrocardiogram. Abnormal.

Syndromogenesis and etiology

It is a monogenic genetic disease of autosomal recessive inheritance.

The basic defect is not known and it has not been possible to determine whether hepatomegaly is secondary to constrictive pericarditis.

Literature

Perheentupa, J. et al.: "MULIBREY nanism, an autosomal recessive syndrome with pericardial constriction." Lancet, II: 351-355.1973.

CONGENITAL NEPHROSIS SYNDROME

Synonymy

S. Of neonatal nephrosis. Nephrotic S. Finnish type". Pediatric microcystic kidney disease.

Syndromography

Clínica

Its onset is intrauterine or during the first month of life and there is a history of prematurity in 20% and always with low weight for their gestational age; voluminous placenta, which
Weight is 25% of normal; frequent toxemia in pregnancy.

Superimposed infections are the main cause of death.

Physical examination. Facies: peculiar with soft ears. Huge edema already at birth or a few days later. Abdomen bulging and tense due to ascites and meteorism, which cause opisthotonus posture and hernias. Skeleton: wide cranial sutures, calcaneo-valgus feet.

Paraclinical examinations

Urine. Massive proteinuria.

Blood. Hypoproteinemia with hypoalbuminemia and hypogammaglobulinemia, increased cholesterol and lipids.

Kidney biopsy. Microcystic dilation of the proximal tubules. Less striking glomerular alterations.

Alphafeto protein. In blood or amniotic fluid to suspect the syndrome in the prenatal period.

Syndromogenesis and etiology

It is transmitted in an autosomal recessive form, with frequent consanguinity in the parents, this family form constitutes the "Finnish type." In some rare cases there is no demonstrable hereditary transmission. It is considered to be due to the passage through the placenta of antibodies produced from a previous pregnancy with stillbirth.

They generally die in the first year of life and it is rare for them to exceed 3 years of age. Kidney transplantation is currently being tested.

The etiology is genetic.

Literature

Ashby, H.: "A case of nephritis in a newly born infant." Soc. Study Dis Child., 1:129, 1901.

Hallman, N. and L. Hjelt: "Congenital nephrotic syndrome." J. Pediat., 55:152, 1959.

Royer, P; R. Habib, H. Mathieu and M. Broyer: Pediatric Nephrology. (Spanish trans.). Ed. Toray, Barcelona, 1975. p. 32.

RECURRENT BRACHIAL NEURITIS SYNDROME

Syndromnography

Clinic

Facies: asymmetrical, with hypotelorism, enophthalmia, sometimes epicanthal folds, and Mongolian inclination of clefts

eyelids, microstomia and cleft palate. Neuritis: of the brachial plexus, of sudden onset, with pain in the shoulder (preferably right), partially disabling that radiates to the hand;
weakness, hyporeflexia and hypoanesthesia of the arm; temporary muscle atrophy and winged scapula; Sometimes the lower cranial nerves (hoarseness) and/or the sympathetic nerves are involved. Growth is limited. Neuritic crises have recurrences at intervals of weeks or years.

Paraclinical examinations

Cerebrospinal fluid and electroencephalogram. Norevils.

Syndromogenesis and etiology

It is assumed that there is a genetic susceptibility of the nerves to hyperergic phenomena. The nerves present “sausage” bulges of the myelin sheaths, with extensive demyelination and remyelination.

It is transmitted by autosomal dominant inheritance, of great penetrance.

The etiology is genetic.

Literature

Drechsfeld, J.: "On some of the rarer forms of muscular atrophes." Brain, 9:578, 1887.
Erickson, A.: "Hereditary syndrome consisting in recurrent attacks resembling brachial plexus neuritis facial features and cleft palate." Paediat Act. Scand., 63:885, 1974.
Guillozett, N. and R. Mercer: "Hereditary-recurrent brachial neuropathy." Am.J. Child., 125:884, 1973.
Taylor, R.A.: "Heredofamilial mononeuritis with brachial predilection." Brain, 83:113, 1960.

NEONATAL NEUTROPHILIA SYNDROME

Synonymy

S. of isoimmune neonatal neutropenia. S. of transient neonatal neutropenia.

Syndromography

Clinic

Newborn with moderate recurrent infections or fulminant infection.

The recommended therapy is with antibiotics if there is infection and steroids or ACTH; The prognosis is generally good with full recovery except in fulminant cases.

Paraclinical examinations

Blood. Leukopenia with severe neutropenia accompanied by macrocytosis. No anemia or thrombocytopenia. Maternal blood contains agglutinins against neutrophils.

Bone marrow rich in the myeloid series, but mature neutrophils are few or absent.

Syndromogenesis and etiology

Passage in the fetus of antibodies against the leukocytes produced by the mother. The phenomenon of passage of these antibodies is quite common, while the effect on the number of neutrophils and clinical manifestations are very rare.

Literature

Hitzig, W.H. andR. Gitzelmann: “Transplacental transfer of leukocyte agglutinins.” Vox Sang. (N.S.), 4:445-456, 1959.

LINEAR SEBACEOUS NEVUS SYNDROME

Synonymy

S. of epidermal nevus. Jadassohn's nevus sebaceus.

Syndromography

Clinic

Nevus sebaceous generally presents from birth, orange-yellow in color, contains deficiency or papillomatous excess of epidermal elements, especially sebaceous glands and immature hair follicles. Over time the lesion becomes verrucous, hyperpigmented and hyperkeratotic;
It is located in the mid-facial region, commonly on the forehead, and extends to the nasal area with a tendency to a linear distribution.
It can affect the trunk and limbs. It is accompanied by variable mental retardation, seizures that vary from sob spasms, myoclonus to grand mal epilepsy.

Other occasional abnormalities are located in the eyes:
lipodermoid of conjunctiva, corneal opacity, colobomas; in the central nervous system, such as cortical atrophy hydrocephalus or in the kidneys such as hamartomas and nephroblastoma. Patients may also present areas of alopecia and cardiovascular malformations, as well as hypoplasia of the teeth.

Paraclinical examinations

Brain ultrasound or computed axial tomography. Ventricular dilations and/or focal cortical atrophies are observed.

Electroencephalogram. Abnormal (spikes and polyspikes).

Syndromogenesis and etiology

This syndrome has a genetic etiology and autosomal dominant inheritance has been suggested. The presence of epidermal nevi and central nervous system abnormalities must have a common origin as a neuroectodermal defect.

Literature

Zaremk, J.: “Jadassohn's naevus phakomatosis: Study based on a review of 37 cases.” J. Ment. Deficiency Res., 22:103-123, 1978.

NEW-CHILDREY SYNDROME

Synonymy

S. of stridor-myelomeningocele.

Syndromography

Clínica

Its onset is neonatal, with congenital stridor and myelomeningocele. The stridor is inspiratory, with a clear voice and a tendency to critical exacerbations. Apneic crises (66%) with cyanosis and severe upper respiratory obstruction.

Seizures (25%). Aspiration due to dysphagia, and, occasionally, pneumonia as a complication.

It is frequently associated with Arnold-Chiari syndrome (66%): myelomeningocele (sometimes occipital) and hydrocephalus, which appears about 3 weeks after surgical correction of the myelomeningocele.

Syndromogenesis and etiology

There is a dysgenesis of the nuclei of the last cranial nerves. Stridor is caused by bilateral paralysis of

the vocal cords, triggered by increased intracranial pressure.

It is transmitted by sex-linked recessive inheritance, affecting the men.

The etiology is genetic.

Literature

Fitzsimon, J.S.: "Laryngeal stridor and respiratory obstruction associated with meningomyelocele." Arch. Dis. Child., 40:687, 1965.

Hollinger, P.C.; T.J. Reichert and P.H. Holinger: "Respiratory obstruction and apnea in infants with bilateral abductor vocal cord paralysis meningomyelocele, hydrocephaly and Arnold- -Chiari malformation." J. Pediat., 92:368, 1978.

New. G.B. and J.A. Childrey: "Paralysis of the vocal cords." Arch.Otolaryngol., 16:143, 1932.

NOONAN SYNDROME

Synonymy

Turner-like syndrome. Male Turner. Turner with normal karyotype.

Syndromography

Clinic

This syndrome is characterized by short stature with prenatal onset in 50% of cases, and mental retardation in 25%.
Facial dysmorphism: antimongoloid deviation of the eyelid fissures and eyelid ptosis, hypertelorism and epicanthus, strabismus.

Low set of hair on the nape of the neck, short and membranous neck with pterygium colli. Shield chest with pectus carinatum or excavatus, valgus position, abnormalities of the vertebral column. Pulmonary valve stenosis, pulmonary branch stenosis. Small penis, cryptorchidism.

Occasionally there may be sensorineural deafness. It affects both the male and female sex. Coagulation defects with partial deficiency of factors XI:C, XII:C and VII:C.

Physical examination. Short stature, winged neck, pulmonary artery stenosis.

Paraclinical examinations

Karyotype. Normal.

Radiology. The described abnormalities are checked.

Echocardiogram. For the study of car defects diovascular.

Coagulogram. Evidence of coagulation defects injured

Syndromogenesis and etiology

It generally occurs sporadically in families and autosomal dominant inheritance has also been described. The coagulation defects found in Noonan syndrome suggest the existence of regulatory factors that control the intrinsic coagulation system, that these factors are under genetic control, and that abnormalities of this regulation occur in Noonan syndrome.

On the other hand, fetal syndrome due to Primodone simulates the Noonan phenotype.

Literature

Sharland, M. et al.: "Coagulation-factor deficiencies and abnormal bleeding in Noonan's syndrome." Lancet, 339: 19-21, 1992.

0CULODENTODIGITAL SYNDROME

Synonymy

ODD syndrome.

Syndromography

Clinic

The clinical delineation of the syndrome highlights Ocular defects: microphthalmia, microcornea, thin and porous iris and short palpebral fissures. The nose is thin with hypoplastic nasal wings; dental hypoplasia; thin, sparse, slow-growing hair.

These patients present syndactyly of the fourth and fifth fingers or third and fourth fingers with camptodactyly of the fifth finger on their hands and feet. They may present neurological dysfunction including hyperactive deep tendon reflexes, ataxia and dysarthria. They do not appear to suffer from mental retardation.

Paraclinical examinations

Radiology. Hypoplasia or aplasia of middle phalanges of one or more fingers and toes. Wide tubular bones and wide alveolar rings.

Brain nuclear magnetic resonance. Demonstrates white matter abnormalities.

Syndromogenesis and etiology

It is a syndrome of monogenic genetic etiology of autosomal dominant inheritance. Many cases represent new mutations.

Variation in gene expression has been described. Healthy consanguineous parents have been reported in one case, also suggesting possible autosomal recessive inheritance.

Literature

Patton, M.A.: "OculodentoOseus syndrome." J. Med. Genet.. 22:386- -389. 1985.

OROFACIODIGITAL SYNDROMES (OFD)

Syndromography

Clinic

Currently, great heterogeneity has been revealed between oro-facio-digital syndromes. In all of them, to a greater or lesser degree, there are oral, facial and digital anomalies so that a clinical classification has been made on the occurrence of unusual anomalies in the affected cases:

OFD TPO I. PAPILLON-LÉA GE-PSAUME SNDROME

It is one of the most delineated, the facies is characterized by frontal bulging, dystopia cantorum, thin nose, cleft pseudolip in the midline of the upper lip. Milio on ears and face. Oral defects: clefts, hyperplastic frenums, pseudo-clefts, cleft palate, asymmetrically cleft soft palate, thick fibrous tracts, lobed tongue, hamartomatous masses, malposition of teeth, supernumerary teeth, small jaw. Digital defects:

syndactyly, clinodactyly and brachydactyly of second and toes

fifth of the hands, in the feet polysyndactyly, syndactyly and brachydactyly, there may be bilateral polydactyly. Central nervous system: slight mental retardation (40%), hydrocephalus, hydranencephaly, porencephaly, agenesis of the corpus callosum. Renal: asymptomatic polycystic kidneys. Other anomalies: precocious puberty, decreased hearing.

OFD TYPE II. MOHR SYNDROME

Facial: characterized by a cleft in the midline, hypertelorism and micrognathia, low-set or rotated ears. Oral: cleft tongue is a constant characteristic (ankyloglossia), aberrant frenulums are less frequent than in type I. Digital: bilateral hexadactyly and polysyndactyly, although it may be asymmetrical. Central nervous system: mental retardation, not constant; microcephaly, porencephaly, internal hydrocephalus, deafness, choroidal coloboma, hypotonia and poor coordination. Respiratory infections.

OFD TYPE IIL. SUGARMAN SYNDROME

Facies: frontal bulging, hypertelorism, anti-mongoloid deviation of the palpebral fissures, apparently low-set ears. Oral: hamartomas and lobed tongue, dental anomalies (malocclusion and extra fingers), bifida uvula, normal palate. Digital: postaxial hexadactyly of hands and feet. Central nervous system: profound mental retardation, strabismus, winking eyes. Pectus excavatum.

OFD TYPE IV. OFD SYNDROME WITH TIBIA DYSPLASIA

Facies: thick nasal root and tip, hypertelorism or telecanthus, micrognathia. Oral: cleft lip, cleft palate, bifid uvula, cleft or hypoplastic maxilla, oral frenulum, hamartomas of the tongue, supernumerary teeth. Digital: preaxial and postaxial polydactyly, syndactyly, clinodactyly, brachydactyly. Dysplastic tibiae, short with bulging, forearms may be shortened, height below the third percentile. Other anomalies:
hypoplastic or absent epiglottis. Pectus carinatum or excavatum.

OFD TYPE V. THURSTON SYNDROME

Facies: middle cleft of the upper lip. Oral: duplicated upper middle frenulum. Digital: heptodactyly, hexadactyly, polydactyly of hands and feet.

OFD TYPE VI. VÁRADI SYNDROME

Facies: hypertelorism, epicanthus, thick nasal tip, cleft lip. Oral: lingual or sublingual intraoral frenums, high or cleft palate, enamel hypoplasia. Digital: postaxial polydactyly, clinodactyly and syndactyly in hands and feet, a constant anomaly is the presence of the central metacarpal in the shape of a fork. Central nervous system: various degrees of cerebellar defects, absence or hypoplasia of the vermis; Dandy-Walker malformation, motor incoordination and mental retardation or absence of language. Other anomalies: arrhinencephaly, deafness.
Cardiovascular: heart defects, aortic stenosis, common atrioventricular canal, coarctation of the aorta. Hypogonadism with micropenis.

OFD TYPE VII

Facies: hypertelorism with pseudocleft of the upper lip. Oral: arched palate with forked tongue, frenulums. Digital:
clinodactyly. kidney: hydronephrosis.

OFD TYPE VIII

Facies: middle cleft of the upper lip, dystopia cantorum, thick and bifid nasal tip. Oral: lobed tongue and hamartomas. Digital: bilateral preaxial and postaxial polydactyly of hands and feet, duplication of the hallux, shortening of the tibia and radius, short stature. Recurrent pneumonia, developmental delay.

Paraclinical examinations

Radiology (bone survey). Look for bone anomalies that can differentiate some types from others.

Abdominal ultrasound. Looking for malformations
kidneys.

Computerized axial tomography of the brain. Due to brain malformations of some types that allow diagnosis.

Echocardiogram. Due to the congenital heart diseases that distinguish type VI.

Syndromogenesis and etiology

OFD syndromes are of genetic etiology with variation in expressivity. Each one has a specific type of inheritance:

OFD type I. X-linked dominant inheritance. Limited to the female sex. Lethal in men.
OFD type II. Autosomal recessive inheritance. Consanguinity has been demonstrated.
OFD type III. Probable autosomal recessive inheritance. Few cases reported.
OFD type IV. Autosomal recessive inheritance.
OFD type V. Autosomal recessive inheritance.
OFD type VI. Possible autosomal recessive inheritance.
OFD type VII. X-linked or autosomal dominant inheritance.
OFD type VII. X-linked inheritance with light expression in heterozygous women.

The basic defect is unknown in all cases.

Literature

Gorlin, R.J.: M.M. Cohen and L.S. Levin: Syndromes of the head and neck 3rd ed. Oxford monographs on medical genetics No. 19. Oxford University Press. New York, 1990 p.676-686.

OSTEOGENESIS IMPERFECTA SYNDROME

Synonymy

Dighton-Adair S. S. of osteosatyrosis. S. of Löbstein. S. of bone brittle. S. de Van der Hoeve.

Syndromography

Clinic

This syndrome is defined by bone fragility that can be accompanied by a reduced life expectancy. Presents low

height, bone deformities, dentinogenesis imperfecta, deafness, blue sclerae and other connective tissue abnormalities.
Taking into account the variation in the clinical manifestations presented and the advances in biochemical and molecular studies, four major groups have been identified: type I, type II, type III and type IV.

The most relevant clinical manifestations of each type are:

Type I. It has a more benign course, fractures are few or no more than 50, generally of long bones and before puberty; It does not affect size, individuals have blue sclerae, normal teeth. There are rarely fractures in the uterus or in the perinatal period, except when the child begins to walk. It has been suggested that type I could be subdivided into IA and IB based on the absence or presence of dentinogenesis imperfecta.

Type II. It is known as a lethal perinatal form. It affects 1 in 20, 000 and 1 in 60,000 newborns. Prematurity and low weight are common. Newborns have short limbs and adopt a frog-like position. The thorax is small, they have a characteristic facial appearance with very blue sclerae, a pointed nose and an extremely soft skull.

Type III. This type is very severe due to bone fragility, muscle weakness and joint contractures. Due to their inactivity, they develop progressive scoliosis that can be so severe that it compromises cardiorespiratory function. They may have hearing loss, dentinogenesis imperfecta and very short stature. The blue tint of the sclera varies with age.

Type IV. In this type, dentinogenesis imperfecta is common, hearing loss occurs in some families. Fractures can be present at birth and even occur in utero or during childbirth. Sometimes fractures of different types are not detected by radiology because they separate adequately or are manifested by femoral bulging.

Growth is frequently normal but height may drop to values below the fifth percentile. Fractures are common in the first month, they can increase when the child begins to walk and do not stop until adulthood, being rare between 20 and 40 years of age. Sclerae are normal, bone deformities are variable, dentinogenesis imperfecta is common, and deafness is observed in some families.

Paraclinical examinations

Radiology (bone survey). For the detection of fractures and bone fragility that characterizes the disease in all its types. Translucent bones, thin diaphyses, deformities and consequences of accidents and fractures.

Blood. Calcium, phosphorus and alkaline phosphatase have normal values and this facilitates the diagnosis of bone metabolism defects.

Ultrasound. During pregnancy it can be useful for the fetal diagnosis of fractures and disproportions due to fractures of the limbs.

Syndromogenesis and etiology

The etiology of osteogenesis imperfecta is monogenic genetics. The basic defect is known in all types. The mutations affect one of the two genes: COL 1"1 and COL 12 that encode type I collagen chains.

Genes affected by mutations produce an abnormal proportion of collagen molecules and this is reflected in the phenotype. For each of the four types of osteogenesis imperfect there are three or more mutations, some are more common and most turn out to be rare mutations.

Type I (autosomal dominant inheritance) presents a common mutation that is non-functional, since there is a shift in the reading frame that gives rise to a large extension of the carboxy-terminal propeptide. In type II (autosomal dominant or recessive inheritance) there are substitutions for the glycyl residue in the domain of the triple helix of the al(I) chains and in type III (autosomal dominant inheritance), it presents a mutation similar to that of type II and in type IV, of autosomal dominant inheritance, the mutation also affects the glycyl residues in domains of the triple helix of the al1(1) and ± (I) chains.

Literature

Byers, P.H.: Osteogenesis imperfecta in Royce P.M. and B. Steinmann: Connective Tissue and Its Heritable Disorders. 1992. Wiley-Less, pp. 317-349.

Byers, P.H.: Disorder's of collagen biosynthesis and structure. Chap. 134 in Scriver, C.R.; TO THE. Beaudet, W.S. Sly and D. Valle: The metabolic and molecular bases of inherited diseases. vol. III, ed. 7 McGraw-Hill, Inc, 1995, pp 4039-4051.

Van der Hoeve, J. and A. De Kleyn: *Blaue Sclera, Knochenbruchigkeit und Schwerthorigkeit". Arch. Ophth., 95:81-93, 1918.

OSTEOLYSIS SYNDROMES

These syndromes are characterized by the resorption and even complete disappearance of some bones (phantom bones syndrome). According to certain characteristics, several syndromes have been described, but at present only four are accepted, which we will explain independently.

fearfully.

TYPE I OSTEOLYSIS SYNDROME (GORHAM)

Synonymy

S. of massive osteolysis. S. of progressive cryptogenetic osteolysis. S. of missing bone.

Syndromography

Clínica

It affects both sexes. It is installed from childhood to young adults. It is almost constantly one-sided. Focal hemangioma affects one or several contiguous bones including adjacent vertebrae. Adjacent soft tissue is affected.

It is the best known clinical form, it is generally painless unifocal, sometimes it is recognized by a pathological fracture that does not heal, stabilizing, without pain or regeneration. Other times it begins in the carpal and tarsus. In the third decade, deformities of the wrists and ankles are observed, as osteolysis becomes more pronounced.

The process always remains localized

Physical examination. Diffuse muscle atrophy is seen. There may or may not be hemangioma of the skin.

Paraclinical examinations

Radiology. The affected bones take on a "sucked candy" appearance. Massive osteolysis.

Syndromogenesis and etiology

The lesions are: hemangiomas and lymphangiomas of the bone.

More than 50 cases have been published, half of them in under 20 years old.

There is angioma of the bone with osteolysis and bone replacement due to fibrosis.

The etiology is unknown.

Literature

See at the end of type IV osteolysis.

OSTEOLYSIS SYNDROME TYPE II (SCHINZ)

Synonymy

S. of nephropathic osteolysis. S. of essential osteolysis.

Syndromography

Clínica

It begins between &-12 years and is characterized by the square appearance of the digital end of the phalanges. Skin: recurrent ulcers on the fingers and soles of the feet, eliminating sequestrations.
Skeleton: progressive osteolysis of the phalanges with shortening of the fingers and, finally, deformations of the hands and feet.
High blood pressure and symptoms of nephropathy.

Paraclinical examinations

Radiology. Progressive osteolysis of the phalanges, until their total disappearance, then affecting other phalanges and metacarpals, which erode laterally, shrinking and eventually disappearing at one end, with cone-shaped diaphysis;
The carpal and tarsal bones may also be affected.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is genetic.

Literature

See at the end of type IV osteolysis.

TYPE I OSTEOLYSIS SYNDROME

Synonymy

S. of dominant osteolysis.

Syndromography

Clinic

It begins between 2-4 years with arthritis of the wrists and knees, which evolve during childhood. In adults, multifocal ostelysis that affects the carpus and tarsus, bilaterally, with excessive resorption and shrinking of the affected bones.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance.

The etiology is genetic.

Literature

See at the end of Type IV osteolysis.

TYPE IV OSTEOLYSIS SYNDROME

Synonymy

S. of acrosteolysis.

Syndromography

Clinic

It begins between 8-12 years, with recurrent ulcers of the hands and feet. Skeleton: flat fingers and oval nails, with progressive digital shortening and deformities in the hands and feet. Contractures in flexion of knees, hips and elbows.

Syndromogenesis and etiology

In some cases it is transmitted by autosomal recessive inheritance.
and in others dominant.

The etiology is genetic.

Literature

Brown, D.M.: D.S. Bradford: R.J. Gorlin et al.: The acro-osteolysis syndrome: Morphologic and biochemical studies." J. Pediatr., 88:573. 1976.

Gorham, L.W.; A.W. Wright; H.H. Shulta et al.: "Disappearing bones: A rare form of massive osteolysis. Report of two cases one with autopsy findings." Am. J. Med., 17:674, 1954.

Hajdu, M. and R. Kauntze: “Cranio-skeletal dysplasia". Br. J. Radiol., 21:42, 1948.

Jackson, J.B.S.: "Boneless arm." Boston Med. Surg. J., 18:368- -369, 1838.

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Wallis, L.A.; T. Asch and B.W. Maisel: "Diffuse skeletal hemangiomatosis; report of two cases and review of literature." Am. J. Med., 37:545-563, 1964.

POTASSIUM-DEPENDENT PERIODIC PARALYSIS SYNDROME

There are two varieties of this syndrome:

1. With a desire to eat salt.
2. No desire to eat salt.

With a desire to eat salt

Synonymy

S. of pleoconial myopathy.

Syndromography

Clinic

It is established in the first months of birth or in early childhood. It is characterized by a lack of strength in the proximal and symmetrical region of the limbs (floppy babies), with episodes of quadriparesis lasting 2-3 weeks.

There may be trunk ataxia and tremor. The characteristic of these patients is their desire to ingest salt, accompanied by thirst.

intense and abdominal pain when the attack sets in, which begins in the morning when waking up.

Physical examination. Muscular hypotonia, no fasciculations or fibrillation, no sensory disorders. There is tendon hyporeflexia.

Paraclinical examinations

Blood. Normal.

Urine. Normal.

Electromyography. It offers a myopia pattern.

Muscle biopsy. Two types of fibers are seen: one is palely colored and the other dark when hematoxylin eosin is used. The dark fibers contain small vacuoles, which are not colored and are diffusely distributed in the intermyofibril areas. Also found in these areas are a small number of fibers containing granular material. There is a marked increase in the number of mitochondria and fatty infiltration in 20-40% of muscle cells.

Syndromogenesis and etiology

The etiology is unknown.

No desire to eat salt.

Syndromography

Clinic

Both sexes are affected equally. It is installed in the first decade. Patients are asymptomatic between attacks, which can be precipitated by: sleep, rest after exertion, alcohol ingestion, cold, mental stress.

Physical examination. During the attack, flaccid paralysis of all muscles except those of facial expression, chewing, swallowing, language and breathing is observed. In some cases the jaw muscles or a limited group of muscles may be affected. There is hyporeflexia of the affected muscles.

The administration of potassium chloride precipitates the attack.

Paraclinical examinations

Blood. Normal serum potassium during and between attacks.

Muscle biopsy. Vacuolization is observed below the sarcolemma, but there are no degenerative changes.

Syndromogenesis and etiology

Autosomal dominant inheritance is suggested. Three types of periodic paralysis are described: type I (hypokalemic); type II (hyperkalemic); type III (normokalemic). In all cases the etiology appears to be genetic.

Literature

Poskauzer, D.C. and D.N.S. Kerr: "A third type of periodic paralysis with normokalemia and favorable response to sodium chloride." Amer. J. Med., 31:328-342, 1961.

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Tyler, F.H., FE. Stephens, F.D. Gunn et al.: "Studies in disorders of muscles VII. clinical manifestations and inheritance of a type of periodic paralysis without hypotassemia." J. Clin. Invest., 30:492-502, 1951.

PARROT SYNDROME

Synonymy

S. Parrot pseudoparalysis. S. de Wegner-Bedanar-Parrot. Parrot's syphilitic osteochondritis. S. of fetal chondrodystrophy. S. of achondroplasia.

Syndromography

Clínic

It is the most common of the chondrodysplasias, it has a frequency of 1:26,000.

Low size from prenatal onset, the length in the newborn is 47.7 cm in the boy and 47.2 in the girl.

The adult size is 13lq 5.6 cm and 124q 5.9 cm, respectively.

It usually occurs in the first 3 weeks of life, rarely after 3 months, and the upper limbs are most frequently affected. It occurs in both sexes.

Craniofacial alterations are megaencephaly, short skull base with early closure of the sphenoccipital suture. Very depressed nasal bridge with prominent forehead. Bull dog dog face. Hypoplasia of the middle third of the face, narrow nasal passages and relative mandibular prognathism.

Lumbar lordosis with thoracolumbar kyphosis. Short limbs with rhizomelic shortening and bowing of the legs.

Fingers of the hands in a trident. Moderate hypotonia. Intelligent-normal

There may be secondary hydrocephalus due to narrowing of the foramen magnum, shortening of the Eustachian tubes. Bad position of the teeth.

Physical examination. Pseudoparalysis and periarticular edema are demonstrated, in addition to the physical characteristics indicated.

Paraclinical examinations

Radiology. There is widening of the joint space, irregularity of the epiphyseal line, thickening of the periosteum and decalcification of the bone. Shortening of long bones, especially humerus and femur, lengthening of the fibula.

Vertebrae: excessive separation of the ossification centers. Small, cuboidal vertebral bodies with shortening of the pedicles, progressive narrowing of the interpedicular distances at the lumbar level.

Pelvis: shortening of the branch of the iliac bone and changes in the sacroiliac curve.

Skull: elongated; moderate or significant dilation of the cerebral ventricles. Occipitalization of the first cervical vertebra, small foramen magnum.

There may be other bone manifestations.

Brain ultrasound at birth. It should also be performed at 4 and 6 months to establish the size of the ventricles.

Syndromogenesis and etiology

Genetic etiology, monogenic with autosomal dominant inheritance. High mutation rate of 1.9• 10 per generation.

Advanced paternal age contributes to new mutations.

Histological evaluation of the epiphyseal line has shown shortening and loss of alignment of the cellular columns and some cartilage cells show mucinoid degeneration.

Prenatal diagnosis during the second trimester of pregnancy can be made by femoral length.

The basic defect is unknown.

Literature

Filly, R.A. et al.: "Short limbed dwarfism: ultrasonographic diagnosis by measurement of fetal femoral length." Ultrasound, 138:653-656, 1981.

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Murdock J.L. et al.: "Achondroplasia a genetic and statistical survey." Ann. Hum. Genet., 33:227-244. 1970.

Parrot, J.M.: "Sur un pseudo-paralysis causee par un altération du systeme osseux chez les nouveaux-nés atteints de syphilis héréditaire." Arch. De Physiol. Norm. Et Path. Paris, 4:319- -333, 470-490, 612-623. 1871-1872.

Parrot, J.M. : "Sur la malformation achondrolasique et le Dieu Phtah." Bull. Soc. d'Anthrop., De Paris 1(3rd series):296-308, 1878.

Silverman, F.N. and S. Brunner: "Errors in the diagnosis of achondroplasia." Radiol Act. (Diag.) (Stockholm), 6:305- -312, 1967.

US Department of Health, Education and Welfare. National Communicable Disease Center, Venereal Disease Program: Syphilis synopsis. Washington, D.C., U.S. Government Printing Office, 1967.

PASSWELL SYNDROME

Synonymy

S. of ichthyosis-nanism-oligophrenia-nephropathy.

Syndromography

Clinic

This syndrome was identified by Passwell et al. (1973) in three children of a consanguineous marriage.

From birth the skin appears erythematous with fine scales, and becomes atrophic in patches, parchment-like on the back of the hands and feet. Small bullae have also been observed. Other symptoms are mental retardation, short height below the third percentile, and renal manifestations that are sometimes only evidenced by the presence of generalized aminoaciduria; Over time these patients suffer from hypertensive pyelonephritis.

Progressive spastic paralysis of the lower limbs, hypergammaglobulinemia and epilepsy have also been reported. Nails and hair are normal.

Paraclinical examinations

Skin biopsy. Hyperkeratosis, vacuolization of the basement membrane and atrophy of the epidermis, lymphocytic infiltration and degeneration of collagen in the dermis.

Urine metabolic tests. General aminoaciduria lized.

Electroencephalogram. Diagnosis of epilepsy.

Protein electrophoresis.

Syndromogenesis and etiology

A monogenic genetic etiology with autosomal recessive inheritance has been proposed. The basic defect and pleiotropic effect of the gene are not known.

Literature

Passwell, J; L. Zipperkowski; D. Katznelson et al.: *A syndrome characterized by congenital ichthyosis with atrophy, mental retardation, dwarfism, and generalized aminoaciduria." J. Pediatr., 82:466-471, 1973.

PENA-SHOKEIR I SYNDROME

Synonymy

S. of ankylosis-facial anomalies-pulmonary hypoplasia.

Syndromography

Clinic

The birth is often premature, the newborn is small for gestational age. 30% are stillborn and those born alive die from complications of pulmonary hypoplasia in the first months of life.

Patients with this syndrome have growth deficiency of prenatal onset with a specific craniofacial dysmorphism in which prominent eyes, hypertelorism, telecanthus, epicanthus stand out; ears with posterior rotation, angled, small, with defects of the normal configuration; nose with a depressed tip, small mouth, high palate, micrognathia, multiple ankylosis of the elbows, knees, hips and ankles; camptodactyly, clubfeet varus, absence of flexion folds and poorly marked dermal ridges. Lung hypoplasia, cryptorchidism.

Polyhydramnio, small placenta, short umbilical cord.

Paraclinical examinations

Brain ultrasound. Brain abnormalities.

Karyotype. Normal (to rule out trisomy 18).

Chest radiology. Lung hypoplasia.

Syndromogenesis and etiology

A monogenic etiology with autosomal recessive inheritance has been proposed in half of the published cases, although the pathogenetic mechanisms are heterogeneous (neurogenic atrophy, abnormal histology of the spinal cord, brain abnormalities), it is proposed that the defects are caused by a decrease in fetal activity: swallowing defects (polyhydramnios); neuromuscular deficiency of the function of the diaphragm and intercostal muscles (produce pulmonary hypoplasia); lack of fetal motility (sequence of fetal akinesia or hypokinesia).

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Hall, J.G.: "Invited editorial comment'. Am. J. Med. Genet. 25:99, ii 986.

PENA-SHOKEIR I SYNDROME

Synonymy

S. cerebro-oculo-facio-skeletal. S. COFS.

Syndromography

Clínica

They are small children for gestational age. They progress with difficulty. There is progressive postnatal growth deficiency and progressive demyelination of the central nervous system. They do not survive 4 years.

They present microcephaly, prominent nasal root, large ears, microphthalmia and blepharophimosis. Small chin, short neck.

A constant fact is the musculoskeletal manifestations: kyphosis, and/or scoliosis, campodactyly with overlapping of the

fingers, flexion contractures especially elbows and knees.
Prominent foot, hip dislocation. 70% may present cataracts.

Paraclinical examinations

Radiology. Acetabular dysplasia, coxa valga, osteoporosis, hypoplasia of the second cuneiform bone gives the appearance of a second metatarsal with a proximal location.

Intracranial calcifications.

Ultrasound and/or computed axial tomography of the brain. Hypoplasia or agenesis of the corpus callosum, cerebellar hypoplasia and intracranial calcifications.

Syndromogenesis and etiology

It is a genetic syndrome in which a history of affected siblings and consanguineous parents shows autosomal recessive inheritance.

The basic defect is not known.

Literature

Pena, S.D.J., et al.: "COFS syndrome revisited." Birth Defects, 14(6B): 205-213, 1978.

PICNODYSOSTOSIS SYNDROME

Syndromography

Clinic

They are short patients with frontal and occipital prominence and delayed closure of the sutures, with persistence of the anterior fontanelle. Facial hypoplasia with prominent nose, high-arched and furrowed palate. Due to the persistence of deciduous teeth, double rows of teeth, delayed eruption and cavities can be observed.

They may have significant pectus excavatum with little breast development, narrow shoulders, kyphosis, scoliosis, increased lumbar lordosis. Short, wide hands with bulbous fingers and wrinkled skin on the back of the hands, flattened, hypoplastic nails.

Two thirds of the patients have had fractures in the jaw, clavicles, and lower limbs. Cases with mental retardation have been reported.

Paraclinical examinations

Radiology (bone survey). Osteosclerosis with a tendency to transverse fractures. Delayed suture closure, persistence of the anterior fontanelle, wormian bones, loss of frontal sinuses, small jaw with an obtuse angle. Dysplasia or loss of the end of the acromion, acrosteolytic dysplasia of the distal phalanges of the fingers, especially the index finger. The referred defects in the spine.

Syndromogenesis and etiology

An autosomal recessive inheritance has been proposed in the majority of cases, although in one family the possibility of it being linked to the X chromosome was raised.

The nature of the basic defect is unknown. The French painter Toulouse-Lautrec has been considered to have this disease.

Literature

Sedano, H.O.; R.J. Gorlin and V.E. Anderson: Pycnodysostosis. Clinical and genetic considerations." Am. J. Dis. Child., 16:70-77, 1968.

WRINKLED SKIN SYNDROME

Synonymy

S. of wrinkling without.

Syndromography

Clinic

In this syndrome, thin skin is observed from birth on the abdomen, back of the hands and feet with a decrease in elasticity in these areas and an increase in the number of palmar and plantar folds. Other features include a prominent venous pattern in the chest, hypotonia, short stature, kyphoscoliosis, winged scapulae, and occasionally mental retardation, myopia, and chorioretinitis. Atrial septal aneurysm.

Paraclinical examinations

Skin biopsy. Elastic fiber abnormalities.

Echocardiogram. Rule out atrial septal aneurysm.

Syndromogenesis and etiology

It presents genetic etiology with autosomal inheritance recessive.

The basic defect must be in correspondence with the synthesis of the elastic fibers.

Literature

Hurvitz, S.A.; A. Baumgarten and R.M. Goodman: "The wrinkling skin syndrome: a report of a case and review of the literature." Clin. Genet.. 38: 307-313., 1990.

POLAND SYNDROME

Synonymy

Anomaly S. Poland. Poland malformative sequence S. S. of pectoral aplasia-syndactyly.

Syndromography

Clinic

An incidence of 1 in 20,000 is reported, and it has been estimated that 10% of patients with syndactyly of the hand have the Poland anomaly. It is more common in males and affects the right side in 75%. It is a unilateral defect that is characterized by absence or hypoplasia of the pectoralis major muscle, hypoplasia of the nipple and areola with asymmetry of position with respect to the normal side.

Patients do not report any symptoms, only that they cannot raise the arm from the affected side to the other side of the chest. In the hand on that side, distal hypoplasia is observed with various degrees of syndactyly, brachydactyly, oligodactyly, and occasionally more severe reduction defects. Occasionally there are hemivertebrae, rib defects, renal anomalies, and Sprengel's anomaly.

Physical examination. Syndactyly associated with an ipsilateral absence of the sternal portion of the pectoralis major. Breast asymmetry in women. In one case, the absence of the nipple was reported.

Paraclinical examinations

Radiology of the chest, spine and hand. HE

They verify the clinical defects already described.

Syndromogenesis and etiology

Its Syndromogenesis is related to a blood flow defect, due to a primary defect of the subclavian artery, towards the distal end of the limb and the pectoral region, so it is proposed that it is a vascular disruptive sequence.

The degree of obstruction determines the severity. The reported family character could explain a certain predisposition to the proposed embryonic mechanism.

The etiology is unknown.

Literature

Adams, R.D.: D. Denny-Brown and C.M. Pearson: Diseases of the muscle. New York, Harper-Row, 1962.

Bovinck, J.N.B. and D.D, Weaver: *Subclavian artery supply disruption sequence: Hypothesis of a vascular etiology for Poland".

Klippel-Feil and Möebius anomalies". Am. J. Med. Genet., 23:903-909, 1986.

Clarkson, P.: “Poland's syndactyly.” Guy Hosp. Rep., 111:335- -346, 1962.

Fraser, E.C.: GM. Romen and E. O'Leary: Pectoralis major defetc and Poland sequence in second cousins: extension of the Poland sequence spectrum". Am. J. Med. Genet., 33: 468-470, 1989.

Mace, J.W. et al.: "Poland's syndrome". Clin. Pediatrician (Phila), 11:98-102, 1972.

Morley, E.B.: "Congenital defect of pectoralis muscles." Lancet, 1:1101-1102, 1923.

Poland, A.: "Deficiency of the pectoral muscles." Guy Hosp. Rep.,6:191-193, 1841.

Skoog, T.: “Syndactyly: clinical report on repair.” Chir Act. Scand., 130:537-549, 1965.

NEONATAL POLYCYTHEMIA SYNDROME

Synonymy

S. of neonatal pletoria. S. of erythrocytosis in children. S. of neonatal polyglobulia.

Syndromography

Clinic

Polyglobulia is considered to be an increase in the number of erythrocytes per unit of plasma volume, accompanied by an increase in the hematocrit value. In order to classify a newborn as polyglobulic, the (venous) hematocrit must be greater than 60%, the number of red blood cells greater than 6 million and the hemoglobin greater than 20 g%. Around the third day of birth, cyanosis appears (40% of cases) of variable intensity, predominantly in the perioral location, although it can be generalized. Other times redness predominates, giving the appearance of plethoric newborns or presenting jaundice secondary to hemolysis.

There may be petechiae and ecchymosis on the skin accompanied by or without mild respiratory distress with polypnea and apnea attacks.

Sometimes there is cardiomegaly due to right overload, mainly with tachycardia. It can lead to heart failure.

Neurological signs are of high incidence, from obtundation to coma, even seizures. Sometimes there are alterations in muscle tone.

Circulatory slowing can trigger renal venous thrombosis and in some cases disseminated intravascular coagulation syndrome.

Hepatosplenomegaly and biochemical alterations such as hypoglycemia and hypocalcemia are rare.

Paraclinical examinations

Blood. High hemoglobin and hematocrit, presence of nucleated erythrocytes. Fetal hemoglobin level.

If central venous pressure is elevated or maternal-fetal transfusion is suspected, hemoglobin (hbf, hba) studies and determination of IgA or ceruloplasmin will be indicated.

Electrocardiogram. Transient abnormalities and not constants.

Chest radiology. Cardiomegaly.

Urine. Elevated excretion of 17-ketosteroids, dehydroandrosterone and epiandrosterone.

Syndromogenesis and etiology

Different conditions can cause polycythemia.

neonatal:

A. Polycythemia with hypervolemia:
1. Errors in transfusion therapy.
2. Fetal transfusions:
to. Maternal-fetal transfusions.
b. Fetal-fetal transfusion (between twins).
c. Placentofetal transfusion (most frequently due to late ligation of the umbilical cord).

B. Polycythemia without hypervolemia (normovolemic):
1. Physiological polyglobulia of the newborn.
2. Benign polyglobulia family:
3. Polyglobulias linked to chromosomal anomalies. It has been described in some trisomies, especially 18 and 21.
4. Polyglobulia due to hypoxemia:
to. Respiratory: linked to excessive height.
b. Cardiovascular: secondary to congenital heart disease tas.
c. Hematic: due to alterations in oxygen exchange.
d. Polyglobulia in children of diabetic, laxemic or cardiac mothers.
5. Polyglobulia secondary to increased spinal stimulation:
to. Tumors.
b. Iatrogenic.
c. Adrenogenital syndrome.

C. Relative polyglobulias (hypovolemic):
1. Dehydration.

Literature

Chaptal, JR; Jean P. Izarn et al.: "La polyglobulie pathologique néonatale. A propos de cinq observations." Pediatrie, 13:515-525, 1958.

Fouron, JC.: Polycythemie néo-natale". Un. Med. Du Canc., 96:1388-1393, 1968.

Gold, A.P. and A.F. Michael: "Congenital adrenal hyperplasia associated with polycythemia." Pediatrics, 23:727-730, 1959.

POLYDYSPONDYLY SYNDROME

Synonymy

Spondylocostal dysplasia. S. hemivertebral. Costovertebral segmentation anomalies.

Syndromography

Clinic

Low stature due to shortening of the trunk (maximum height 150 cm), kyphoscoliosis; chest defects due to rib malformations.

It may be accompanied by congenital dislocation of the hips, unilateral shortening of limbs and non-bony anomalies such as duplication of the colon, renal or lung aplasia, anal imperforation, alopecia with skin atrophy.

Paraclinical examinations

Radiology. Hemivertebrae, fusions of vertebral bodies or posterior cervical arches; ribs welded or absent; rachischisis, transverse process aplasia.

Abdominal ultrasound. Looking for other malformations described.

Karyotype. To rule out associated chromosomal abnormalities.

Syndromogenesis and etiology

Monogenic genetic etiology, autosomal dominant inheritance. There is another form of spondylocostal dysplasia (Jarcho-Levin syndrome) of autosomal recessive inheritance. The syndrome has been reported in association with translocation of chromosomes 14-15.

The basic defect is not known.

Literature

Lorenz, P. and E. Rupprecht: "Spondylocostal dysostosis: dominant type." Am. J. Med. Genet., 35: 219-221, 1990.

TRANSIENT POLYPNEA SYNDROME OF THE NEWBORN

Syndromography

Clínic

Generally, it is a full-term newborn who has an increased respiratory rate on the first day of birth, some intercostal retraction and moaning; slight cyanosis, no hoarse rales or transmitted noises are heard.

The respiratory rate can remain elevated for 2-5 days.

Paraclinical examinations

Electrocardiogram. Normal.

Chest radiology. Suggestive of vascular congestion. Mild cardiomegaly.

Gasometry. Within normal limits.

Syndromogenesis and etiology

It is suggested that it may represent a failure in the reabsorption of fluid from the lungs.

The prognosis is favorable with the disappearance of the radiological findings between the first and fifth days.

The etiology is unknown.

Literature

Avery, M.E.; 0.B. Gatewood and G. Brumley: "Transient tachypnea of newborn. Possible delayed resorption of fluid at birth." Amer. J. Dis. Child., 111:380-385, 1966.

POMPE SYNDROME

Synonymy

Pompe disease. S. of congenital rhabdomyoma of the heart. Von Gierke's disease. S. glycogenosis type I. S. of acid maltose deficiency. S. of diffuse glycogenic cardiomegaly. S. of generalized idiopathic glycogenosis. S. from generalized glycogenosis to a neuromuscular form. S. of diffuse glucogenic cardiomegaly. S. glycogenosis type Cori II. Glycogen storage system. Van Creveld's hepatorenal glycogenosis syndrome.

S. of hepatonephromegalic glycogenosis.

Syndromography

Clínic

It predominates in men and affects the muscles, heart and nervous system. It appears 2 or 3 months after birth.

This disorder usually manifests itself during the first year of life by symptomatic hypoglycemia or hepatomegaly.

Among the characteristic features are a rounded facial appearance with full cheeks and a prominent abdomen due to large hepatomegaly contrasting with the thinness of the limbs. Accompanying hyperlipidemia can produce eruptive xanthomas and retinal changes typical of lipidemia.

Growth is usually normal in the first months and then it is delayed and adolescence is delayed. Mental development may be normal or delayed in relation to hypoglycemic attacks. This may be accompanied by glycemic levels less than 15 mg/dL.

Four clinical variants are recognized according to the degree of participation of the different organs, although overlapping symptoms and signs may occur:

1. Cardiomegalic variety.
2. Generalized variety.
3. Neuromuscular variety.
4. Infant variety of late acid maltase deficiency.

Varieties 1, 2 and 3: appear in both sexes in the first month of life. There is vomiting, anorexia, drooling, extreme weakness, difficulty in development. Severe mental retardation may be present, frequent respiratory infections, late dyspnea.

Variety 4: the onset is later than in previous varieties. Weakness of the hip muscles, positive Gower signs. Achilles tendon contraction. Firm and elastic muscles, the anal sphincter may be dilated; defective bladder contraction. Mental retardation or normal development.

Physical examination. Weak sucking and movements, weak crying. Macroglossia and exoglossia reminiscent of the facies of cretinism. The muscles feel firm as rubber but there is generalized hypotonia.

Cardiomegaly of variable degree, pointed systolic murmur, progressing to heart failure. No hepatosplenomegaly.

Motor delay, osteotendinous areflexia, sometimes facial paralysis, eyelid ptosis and loss of sphincter control.

In the generalized variety there are various neurological disorders and late cyanosis.

Paraclinical examinations

Muscle biopsy. "Honeycomb" glycogen accumulation, absence of alpha 1,4 glucosides.

Chest radiology. Cardiomegaly.

Prenatal study. Amniotic fluid cell culture.

Blood. Massive glycogen deposition in leukocytes, absence of alpha 1,4-glucosidase activity. Blood glucose, glucose and galactose tolerance test, glucagon and epinephrine normal.

Liver enzymes are practically normal. There is hyperlipidemia that includes cholesterol and triglycerides, which can

be extreme with levels up to 5,000 to 6,000 mg/dl.

Hyperuricemia that can become more serious after adolescence. When there is platelet involvement, hemorrhages occur.

Leukocytes: Absence of alpha 1.4-glucosidase activity.

Electrocardiogram. P wave accentuation. Short PR interval. ST segment elevation, inverted T wave.

Electromyography.

Ultrasound. Shows increased liver size.

Liver biopsy. It makes the exact diagnosis by showing the glucogenic infiltration of the cells with preservation of the architecture. The enzymatic study in it allows us to differentiate the two varieties: Ia and Ib that are indicated in syndromogenesis.

Syndromogenesis and etiology

It is due to an accumulation of glycogen in different tissues due to a lack of alpha 1,4-glucosidase (acid maltase) in the heart, skeletal muscles, tongue, liver and skin. This enzyme hydrolyzes maltose and is essential for converting glycogen into glucose. The structure of glycogen in this syndrome is different from other forms of glycogenosis.

The syndrome is explained by an abnormal storage of glycogen in different organs (liver, kidney, heart, etc.) due to a congenital enzymatic defect in the breakdown of glucose. The defect can be of two types that mark the two varieties:

Type Ia. Glucose 6-phosphatase deficiency.

Type Ib. G6P microsomal translocase deficiency.

The latter, much less common, has the same symptoms but is associated with: neutropenia, limitation of neutrophil migration and recurrent pyogenic infections. It is more serious than the previous one. It is known as pseudotype I.

It is transmitted by autosomal recessive inheritance.

The etiology is genetic.

Literature

Greene, H.L. et al: "Type I Glycogen storage diseases. A Metabolic basis for advances in treatment." Adv. Pediatric, 26: 33, 1979.

Howell, R.R. In: Stanbury-Wyngaarden, Frederickson: Metabolic basis of inherited disease. 3 ed. New York, McGraw-Hill, 1972., p.149.

Huijing, C.F.; S Van Creveld and G. Losekoot: “Diagnosis of generalized glycogen estorage disease (Pompe's disease)”. J. Pediatr., 63:984, 1963.

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Roth, C.J. and H.E. Williams: "The muscular variant of Pompe's disease", J. Pediat., 71:567-573, 1967.

Swaiman, K.F: W.R. Kennedy and H.S. Sauls: Late infantile acid maltase deficiency". Arch. Neurol., 18:642-648, 1968.

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Von Gierke, E.: "Hepato-Nephromegaly Glikogenia: Glykogenspeicherkrantkheit der leber und nieren". Beitr. Path. Anat., 88: 497, 1929.

POST-VACCINATION SYNDROMES

These syndromes constitute the complications of vaccination against smallpox.

Syndromography

Clínic

The following varieties can be considered:

1. Accidental infection. The accidental implantation of the bovine virus in the eyes, lips, in a part of the body in the absence of eczema or other pre-existing skin lesion.
2. Generalized vaccinia. The generalized dissemination of vaccine lesions in the absence of eczema or other preexisting skin lesions.
3. Eczema vaccinatum. The widespread spread of the vaccine lesion or the local implantation of vaccine in a person who has eczema or a past history of eczema. This person may be the patient who has been vaccinated or a contact of someone recently vaccinated.
4. Vaccinia necrosum (Progressive Vaccinia). Spread of necrosis at the vaccination site with or without necrotic metastatic lesions in other parts of the body.
5. Encephalitis. Post-vaccination complication of the central nervous system that includes, separately or in combination, the following symptoms: meningeal signs, ataxia, muscle fatigue, paralysis, lethargy, coma or seizures.
6. Others. Vaccinal lesions complicating skin conditions such as eczema associated with other lesions not mentioned above such as: generalized urticarial reaction, erythema bullous multiforme, secondary bacterial infection.

Literature

Neff, J.M.; J.M. Lane J.H. Pert et al.: "Complications of smallpox vaccination. I. National survey in the United States, 1963*". New Eng. J. Med., 276:125-132, 1967.

Neff, J.M.; RH. Levine; J.M. Lane; et al.: "Complications of smallpox vaccination in the United States, 1963. II. Results obtained by four statewide surveys." Pediatrics, 39:916-923, 1967.

POTTER SYNDROME

Synonymy

S. of bilateral renal agenesis. S. reno-facial.

Syndromography

Clinic

This syndrome is seen in children who have multiple genetic abnormalities. The facies is characteristic: large and soft ears, set low with cartilage alterations, closed eyelids with epicanthus and hypertelorism, long and flat nose, curved "parrot" type, the child has a senile face (Potter's facies). The skin is dehydrated. The kidneys are not palpable and there is no urination.

It is associated with pulmonary hypoplasia and other malformations in the hands and feet with clubbed fingers. Malformation of the thoracic and lumbar spine; genital malformations, hypospadias.

Children who have double renal and ureteral agenesis die at birth in the following 2-3 days. If the agenesis is of only one kidney, it is only discovered accidentally at any age.

Syndromogenesis and etiology

These are congenital malformations, but no hereditary factor has been determined, although it has been reported in two brothers. Trisomy 18 syndrome has many similar characteristics.

The etiology is unknown.

Literature

Passarge, E. and J.J.M. Sutherland: "Potter's syndrome." Amer. J. Dis.Child, 109:80-84, 1965.

Potter. E.L.: "Bilateral renal agenesis." J. Pediat.. 29:68-76. 1946.

PRADER-WILLI SYNDROME

Synonymy

PWCR.S. by Prader-Labhart-Willi.

Syndromography

Clinic

There are no established incidence values, but until 1987 the Prader-Willi syndrome association had a registry of 1,595 affected people in the United States and Canada. Others have estimated an incidence of 1:15,000.

The most important clinical manifestations are: severe hypotonia from birth, insatiable appetite, and obesity that begins between 1 and 3 years of age, especially truncal, which generally develops when hypotonia decreases. Small hands and feet that become evident as the child grows.

During pregnancy there is a reduction in fetal movements.

Genitals: Small penis and cryptorchidism. Hypogona-dysm secondary to hypogonadotropism.

Neurology: Variable mental retardation I.C. between 20 and 80. Poor motor coordination.

Microcephaly: Seizures. Hypothalamic dysfunction. Temperature regulation disorder. Motor development is delayed in the first years, for example they walk around 2 years old. Diction and language problems are common, although the cause has not been clarified, suggesting that it is related to hypotonia of the muscles of the mouth and tongue, or reduced saliva production. Attention by the speech therapist is recommended.

Endocrinology: Diabetes related to the severity of the obesity.

Paraclinical examinations

High resolution karyotype (prometaphasic) and molecular techniques.

Glucose tolerance curve.

Determination of insulin among other studies endocrine.

Syndromogenesis and etiology

An interstitial 15ql1 deletion has been reported in 59.5% of patients; however, there must be chromosomal and non-chromosomal factors, a fact that has led to the proposition that Prader-Willi syndrome is a sequence rather than a syndrome.

15q11 deletion of paternal origin and maternal uniparental dyssonia suggest genomic imprinting as an explanation for the development of this entity.

Literature

Bray, GA. et al: The Prader-Willi syndrome: a study of 40 patients and a review of the literature". Medicine, 62:59-80,1983.

Frins, J.P.: "The Prader-Willi syndrome and the Sotos syndrome: syndromes or sequences? (Letter)". Clin. Genet., 33:457-458, 1988.

PRUNE-BELLY SYNDROME

Synonymy

S. of abdominal muscle agenesis. Sequence of early urethral obstruction.

Syndromography

Clínic

The defects are detected at birth, characterized by a large flaccid belly with rough and thin skin, outlining the morphology of the gastrointestinal package, the greater, lesser and transverse oblique muscles are missing; Of the rectus there is only the supraumbilical portion. In the urinary system, megabladders (three to four times larger than normal), long and dilated ureters, and hydronephrotic or dysplastic kidneys stand out. The male:female ratio is 20:1. It is accompanied by bilateral intra-abdominal cryptorchidism. Varus feet and hip dysplasia.

Other anomalies are: persistence of the urachus, congenital heart disease, unicrognathia, torticollis, omphalocele, incomplete anal atresia, etc.

Prognosis: 20% are stillborn, 30% die before age 2.

Paraclinical examinations

Abdominal ultrasound. Referred malformations.

Echocardiogram. Congenital heart disease.

Syndromogenesis and etiology

The defect begins with a urethral obstruction that gives rise to a sequence of defects that ranges from oligohydromnios and complex deformity, to bladder distension that can continue towards hypertrophy of the bladder wall, hydroureter, abdominal distension with the consequent deficiency of abdominal muscles and excess abdominal skin.

Literature

Pagon, R.A.; D.W. Smith and T.H. Shepard: Urethral obstruction malformation complex: a cause of abdominal muscle deficiency and the 'prune belly'". J. Pediatr., 94: 900-906, 1979.

PSEUDOHURLER SYNDROME

Synonymy

S. of pseudoHurler polydystrophy. S. mucolipidosis type III.

Syndromography

Clinic

In this syndrome, the first clinical manifestations occur around the age of 5 years. There is a decrease in growth, moderate mental deficiency. The facies appears coarse, there may be acne, corneal opacity appears, increased joint volume, aortic regurgitation, inguinal hernia, lumbar lordosis, short neck, carpal tunnel compression.

Paraclinical examinations

Radiology. Spine: platyspondyly; pelvis: widening of the iliac wings; flattening of the femoral epiphysis; generalized osteoporosis.

Slit lamp examination of the cornea. Phase contrast microscopic examination. Phenomenon of cellular inclusions (I-cell).

Syndromogenesis and etiology

Of genetic etiology, autosomal recessive inheritance, the basic defect corresponds to insufficiency of lysosomal enzymes, as in Leroy syndrome.

Literature

Thomas, G.H. et al.: "Mucolipidosis II (pseudoHurler polydystrophy): Multiple lysosomal enzyma abnormalities in serum and cultured fibroblast cells." Pediatrician Res., 7:751-756, 1973.

PSEUDOTALIDOMIDICAL SYNDROME

Synonymy

Phocomelia SC. Hypomelia-hypotrichosis-hemangioma. Phocomelia-flexion deformities-facial anomalies.

Syndromography

Clínic

The acronym SC are the initials of the first families in which this syndrome was observed: family S originating from Germany and Switzerland and family C from Italy and Ireland.

75% of the reported cases present: tetraphocomelia, bilateral cleft lip and palate, ectrodactyly and malposition of the thumb, syndactyly. Low birth weight and deficits in both intrauterine and extrauterine development.

The hair may be sparse and silvery blonde. On the face, in addition to the malformations indicated, there may be hypertelorism and capillary hemangioma, the orbits are small and the eyes prominent, bluish sclerae and malformed ears.

Phocomelia or hypomelia are of variable degrees but almost always symmetrical.

Cryptorchidism and a relatively large penis in relation to body size are reported in the genitals.

Paraclinical examinations

Radiology. Checks and specifies aplastic or hypoplastic bones.

Syndromogenesis and etiology

Of genetic etiology, autosomal recessive inheritance. Implications in the mitotic mechanism have recently been suggested due to a structural protein of the kinetochorus.

Literature

Freeman, M.V.R. et al.: "The Roberts's syndrome." Clin. Genet., 5:1-16. 1974.

Jaks, E.W. et al.: "Studies of mitotic and centromeric abnormalities in Roberts's syndrome: implications for a defect in the mitotic mechanism." Chromosoma, 100: 251-261, 1991.

POPLITEUSEPTERIGION SYNDROME

Synonymy

Flughaut. Popliteal phaciogenitis syndrome.

Syndromography

Clínic

The most striking characteristic of this syndrome is the pterygium (cutaneous membrane) that extends from the heel to the ischial tuberosity, limiting the extension, abduction and rotation of the leg.

It also presents oral abnormalities determined by dimples on the lower lip, bilateral synechiae between the mandible, upper and lower bulla, cleft lip with or without cleft palate.

In the feet there is a peculiarity, the skin that covers the first joint of the foot is shaped like a pyramid with the apex extending towards the nail. Genital anomalies in men that may consist of: cryptorchidism, absent, bifid or ectopic scrotum and inguinal hernia.

In females, absence or displacement of the labia majora, hypertrophy of the clitoris and hypoplasia of the uterus have been observed.

Surgical treatment of pterygium must be carried out with extreme care because it may contain the sciatic nerve and popliteal artery.

Paraclinical examinations

The diagnosis is eminently clinical.

Syndromogenesis and etiology

The etiology is genetic with autosomal dominant inheritance, with wide variation in clinical severity.

Literature

Hall, J.G. et al.: Limb pterygium syndromes: A review and report of eleven patients". Am. J. Med. Genet., 12: 377-409, 1982.

CHERUBISM SYNDROME

Synonymy

Familial mandibular fibrosis. Familial benign giant cellular tumor of the jaw. Multilocular cystic disease of the jaw.

Syndromography

Clinic

The clinical signs that characterize this syndrome begin with an increase in jaw volume around 2 years of age, progressing until 7 years of age and then stabilizing until puberty, decreasing or regressing in the third decade of life.

It has various degrees of deformity: mild, when only the region of the first molar bulges; severe, when the entire jaw thickens including the gums and narrows the palate, in these cases there is bulging of the cheeks, pushing the eyes upward as if "looking towards the sky", which allows the lower part of the sclera to always be seen.

Teeth emerge early but crowd together and fall out early.

Thickened and hard submandibular and upper cervical nodes may appear. Cases have been described with multilocular involvement in the ribs.

Paraclinical examinations

Radiology. Polycystic, foam-like image of the jaw, excluding the condyles. It also affects the upper jaw and sometimes the anterior end of the ribs.

Syndromogenesis and etiology

It has genetic etiology with autosomal dominant inheritance, with variation in gene expressivity.

The basic defect is not known.

Literature

Peters. W.J.N.: "Cherubism: a study of twenty cases from one family." Oral Surg. 47: 307-311, 1979.

RABSON-MENDENHALL SYNDROME

Synonymy

Acanthosis nigricans-insulin-resistant diabetes-facies.

Syndromography

Clinic

Children present early dentition, coarse facies and strong hirsutism, dry skin, acanthosis nigricans, thickened nails, enlarged penis or clitoris, diabetes, cytoacidosis and insulin resistance. There have been reported cases that died in childhood and others have surpassed this age.

Pineal hyperplasia has been found in autopsies.

Paraclinical examinations

Skin biopsy. Abundant glycosaminoglycan (GAG) deposits, mainly hyaluronic acid, in the papillary dermis of acanthosis nigricans lesions. Papillomas, pigmentation and keratinization.

Blood glucose. Hyperglycemia.

Determination of insulin in blood. Hyperinsulinemia.

Syndromogenesis and etiology

It is a genetic defect due to a mutation in the insulin receptor gene, which determines a defect between the synthesis of the receptor and its insertion into the membrane.

Literature

Moncada, V.Y, et al.: "Insulin-receptor biosynthesis in cultured lymphocytes from an insulin-resistant patient (Rabson-Mendenhall syndrome): evidence for defect before insertion of receptor into plasma membrane." Diabetes, 35: 802-807, 1985.

FAMILIAR HYPOPHOSPHATEMIC RICKETS SYNDROME

Synonymy

X-linked hypophosphatemia. Primary hypophosphatemic rickets resistant to vitamin D. Phosphaic diabetes. Vitamin D-resistant rickets.

Syndromography

Clinic

In this type of rickets, children are normal in their growth until serum phosphorus levels fall to low values around 6 months of age. From this moment on, light to moderate growth deficiency begins, until reaching an adult height of 130-160 cm.

Patients present arching of the lower limbs, waddling gait. Delayed tooth eruption, enamel hypoplasia, gingival and periapical infections.

Dolichocephaly, craniosynostosis, pseudofractures, scoliosis. Deformities can progress in adults: exostoses in the bone sites of muscle insertion, fusion of vertebrae and scoliosis and sensorineural deafness.

Paraclinical examinations

Radiology. Bowing deformities of the limbs are noted, especially in the distal region of the femur and proximal region of the tibia in which there is a physis amplitude. Anteroposterior examination revealed rickets changes in the knees. Evidence of hyperparathyroidism is generally absent, unlike VDDR (Vitamin D Dependent Rickets) I and II rickets. With age there is an increase in density. Pseudofractures are radiological evidence of osteomalacia.

Blood. Serum calcium presents levels at the lower limit of normal values. There is hypophosphatemia that must be carefully examined, since in children normally serum pH levels are higher than in adults and are related to their ingestion in food, which is why these determinations must be made on an empty stomach and the patient's age must be taken into account.

Serum potassium and/or bicarbonate levels are normal.

Serum levels of active vitamin D -1.25 (OH)2D- are normal or low. Parathyroid hormone levels are normal. Serum alkaline phosphatase is increased.

Urine. There may be glycosuria.

Syndromogenesis and etiology

The etiology is genetic, with dominant inheritance linked to the

There are still controversies about the expression of the gene as dominant or if it is the result of an overdose effect (due to Lyon's genetic hypothesis of random inactivation of an X chromosome in women).

The current hypothesis of the basic mechanism is supported by a mouse model with great genetic homology in humans (Hyp).
It poses a defect in the villous edge of the renal proximal tubule for pH transport, but there is controversy as to whether this defect is expressed directly in the kidney, bone or intestine. The combined treatment with the active form of vitamin D and phosphorus supplement is currently considered the best, monitoring the treatment with monthly serum determinations of calcium, phosphorus and alkaline phosphatase.

Literature

Whyte, M.P.: Osteopetrosis and the heritable forms of rickets. In. Connective tissue and its heritable disorders molecular genetic and medical aspects (Royce P.M., B. Steinmann. eds.) pp: 572- -575, Wiley Liss, New York, 1993.

VITAMIN D-DEPENDENT RICKETS SYNDROME

Synonymy

Pseudovitamin D-deficient rickets. Familial pseudodeficiency rickets. S. of vitamin D

Syndromography

Clinic

Patients who suffer from this syndrome present a clinical, radiological and histopathological picture similar to rickets due to vitamin D deficiency. However, they do not have nutritional problems.

In 1961 Prader and collaborators described two children with rickets that perfectly simulated nutritional rickets; their organisms reacted only to large doses of vitamin D. The term "vitamin D-dependent" was used to describe this entity.

In 1978, Brooks and other researchers reported a case with a clinical picture similar to that of vitamin D-dependent, but which presented high levels of the active form of vitamin D (1,25 (OH)2 D) and the term "hereditary resistance to 1,25-dihydroxyvitamin D" was used.

Currently, both syndromes are called vitamin D-dependent rickets (VDDR) and are designated as:

VDDR type I and VDDR type II.

Clinical manifestations. Similarities and differences:

VDDR type I. Clinical manifestations of nutritional rickets that begin in the middle of the first year of life, there is muscle weakness and little progress.

VDDR type II. Clinical manifestations similar to VDDR type I with the only difference that 50% of patients present hypotrichosis or complete alopecia that may be obvious at birth or may develop in the first months of life.

Other additional characteristics that clinically differentiate both syndromes are: oligodontia, epidermal cysts and cutaneous milio in type II.

Paraclinical examinations

Blood. Alkaline phosphatase increased, calcium and phosphorus normal or low.

Urine. Renal tubular acidosis, hyperaminoaciduria.

Syndromogenesis and etiology

Genetics: autosomal recessive. An enzyme deficiency prevents the production of sufficient active metabolite from vitamin D; This metabolite may be 1-25-hydroxycholecalciferol, which acts to facilitate intestinal absorption of calcium.

The following defects occur:

1. Absence of the receptor for vitamin D (1.25 (0H) 2 D).

2. Decrease in the binding capacity or affinity to the receptor.

3. Failure of the vitamin D receptor complex to localize to the nucleus.

4. DNA binding defects to the hormone receptor complex.

Theoretically, in VDDR type I, high doses of the active form of vitamin D could be an effective treatment. In both types, treatment with high doses of calciferol and calcium relatively compensate for the resistance in the target tissues.

Literature

Fraser, D. and R.B. Salter: "The diagnosis of various types of rickets." Pediat. Clin. N. Amer., 5:417, 1958.

Maroteaux, P.: Maladies Oseusses de l'Enfant. Ed. Flanmmarion, Paris, 1974.

Prader, A; R. Illig und E. Neierli: "Eine kissonde Form der primaeren vitamin-D-resistant Rachits mit Hypocalcaedemie und autosomal-dominant erbgang die hereditaire Pseudo- -Mangeirachtis." Hello. Paediat. Acta, 16:452. 1961.

Whyte, M.P.: Osteopetrosis and the heritable forms of rickets. In Connective tissue and its heritable disorders, molecular, genetic and medical aspects (Royce P.M., B. Steinmann eds.) pp 570- -572. Wiley Liss, New York, 1993.

REFSUM SYNDROME

Synonymy

Polyneuritiform ataxic heredopathy.

Syndromography

Clinic

It is characterized by the presence of retinitis pigmentosa, ichthyosis, hypertrophic neuropathy with motor and sensory impairment, cerebellar ataxia and sensorineural deafness.

Visual symptoms usually begin after age 20. The neurological ones, in childhood or early adulthood.

Paraclinical examinations

Fibroblast culture. It allows the diagnosis of the portators.

Amniocentesis. Establish prenatal diagnosis.

Blood. Very high levels of phytanic acid are found in plasma that are 100 times higher than in normal cases. Phytanic acid represents 5-20% of the total fatty acids in plasma in affected cases.

Electroretinogram. Abnormal, typical of retinitis pigmentary.

Electroencephalogram. Abnormal in most cases.

Audiometry. Deafness.

Nerve biopsy. Hypertrophic polyneuritis.

Metabolic tests in urine and blood.

Gas chromatography. To demonstrate the presence of acid phytanic do.

Syndromogenesis and etiology

It is a hereditary syndrome in which a metabolic defect is found that consists of an abnormality in the degradation of phytanic acid (a fatty acid with 20 carbon atoms endowed with 4 methyl groups).

These patients do not hydroxylate phytanic acid and therefore cannot transform it into prostanic acid, which is one of the essential steps in the catabolism of phytanic acid, which results in its accumulation in the blood. The lesion of the temporal bone includes degeneration of the vascular stria and atrophy of the organ of Corti. The vestibular system (upper part) is normal. Refsum syndrome is of genetic etiology with autosomnic recessive inheritance.

The basic defect has been identified as a deficiency in the activity of the enzyme peroxisomal-phytanic acid-oxidase that catalyzes the alpha-oxidative process by which phytanic acid is shortened by one carbon atom.

Literature

Steinberg, D. et al.: "Refsum's disease: nature of the enzyme defect." Science, 156:1740-1742, 1967.

Paparella, M.M. and D.A. Shumrick: Otorhinolaryngology. Scientific-Technical, Havana, 1984. pp:1825.

CHILDHOOD POLYCYSTIC KIDNEY SYNDROME

Synonymy

Recessive polycystic kidney disease. Potter's S. Childhood polycystic kidney disease type I. Caroli disease PKD2.

Syndromography

Clinic

Its onset is intrauterine, the child is born with a bulging abdomen and oligohydramnios. Facies: sunken eyes, with groove under lower eyelids; large flaccid ears, set low, micrognathia (Potter's facies). Abdomen: bulging laterally, both large, lobed kidneys can be palpated.

Hepatomegaly sometimes. Respiratory symptoms: pulmonary hypoplasia with pneumomediastinum and respiratory failure. Other associated anomalies: occasionally, liver, neurological, thymus, lungs and seminal vesicles. There is frequently pyloric stenosis, bile duct dilation and liver fibrosis that evolves into portal hypertension, especially in older children.

Paraclinical examinations

Urine. Proteinuria and sometimes hematuria, ultimately uremia.

Radiology. High-dose urography and delayed imaging, with radially elongated clearances throughout the parenchyma, lasting days.

Syndromogenesis and etiology

Kidneys: thickened, spongy, retain their shape but are lobed, covered with numerous 1-2 mm cysts; On section they have a honeycomb appearance, due to radially elongated cysts, without fibrosis or abnormalities of the urinary tract. Liver: cysts due to proliferation of bile ducts (20%) and sometimes periportal fibrosis. Exceptionally, cysts in the pancreas, lung and thymus.

The survival, which is very short, is probably due to the persistence of several generations of nephrons.

In 22.2%, this syndrome is transmitted by autosomal recessive inheritance. Its frequency is double in females. In other cases, the etiology is sporadic and may be due to different causes.

Abnormal expression of the MYC oncogene could play an important role in the pathogenesis of the disease, since high levels of MYC mRNA have been observed in polycystic kidneys.

Literature

Bristowe, F.: "Cystic disease of the liver associated with similar disease of the kidneys." Tr. Path. Soc. London, 7:235, 1856.

Cowley, B.D.; FL. Smarda; JJ. Grantham and J.P. Calvert: "Elevated MYC protoncogene expression in autosomal recessive polycystic kidney disease." Proc. Nat. Acad. Sci., 84:8394-8398, 1987.

Lathrop, D.B.: "Cystic disease of the liver and kidneys." Pediatrics, 24: 251, 1959.

Potter, L.E.: "Facial characteristics of infants with bilateral renal agenesis." Amer. J. Obstetr., 51:885-888, 1946.

Potter, L.E.: "Bilateral renal agenesis." J. Pediatr., 29:68-76, 1946.

JUVENILE POLYCYSTIC KIDNEY SYNDROME

Synonymy

Autosomal dominant polycystic kidney disease. Adult polycystic kidney disease type I. PKDI.

Syndromography

Clinic

It begins between 3-4 years (exceptionally in neonates) and is manifested by hematuria with dysuria, polyuria and nocturnal enuresis. turn, as first symptoms. Continuous, slow lumbar pain.

Physical examination. Abdomen: bulging with palpable kidneys, generally large. Liver: if it is cystic there is hepatomegaly (sometimes also splenomegaly if there is portal hypertension due to fibrosis). Tendency toward hyponatremic dehydration, with polyuria.

It can be associated with aneurysms of the cerebral arteries (10%) and cardiovascular anomalies: coarctation of the aorta and endocardial sclerosis.

Paraclinical examinations

Blood. Anemia.

Urine. Decreased concentration.

Radiology. Descending urography (if kidney function is greater than 50%): abnormalities of the calyxes, with dilation and lengthening.

Kidney ultrasound.

Syndromogenesis and etiology

Thickened kidneys up to 2-5 times the normal size are observed, with intercommunicating cysts of 3-5 cm in diameter, with liquid content, in variable numbers. In 40% of cases, asymptomatic cysts coexist in the liver until periportal fibrosis occurs.

It is considered that there may be a toxic metabolic action that causes secondary kidney injuries.

A great phenotypic heterogeneity in clinical manifestations has been reported. The onset of the condition or kidney cysts varies between families. There are cases of families that were diagnosed as carriers of PKDI through a fetal ultrasound.

The PKDI locus has been located on chromosome 16, specifically on p. The basic defect seems to be related to proteins in the structure of cell membranes.

The hypertension that may accompany this syndrome is due to increased renin release, probably caused by ischemia secondary to renal expansion of the cysts.

The familial aggregation of early clinical forms is suggested by genetic modifications.

The etiology is genetic.

Literature

Chapman, A.B. et al.: "The renin-angiotensin-aldosterone system and autosomal dominant polycystic kidney disease." New. Eng. J. Med. 323:1091-1096, 1970.

Kaplan, B.S.; L. Rabon: M.B. Nogrady et al.: "Autosomal dominant polycystic renal disease in children." J. Pediat., 90:782, 1977.

Lejars, P.P.: Du gros rein polykystique de ládulte. Ed. Steinnheil, Paris, 1888.

Reeders, S.T. et al.: "Regional localization of the autosomal dominant polycystic kidney disease locus." Genomic, 3:100- -155, 1988.

Royer, P.; H. Mathieu and M. Broyer: Pediatric Nephrology (trans. Spanish). Ed. Toray, Barcelona, 1975. p.13.

Zerres, C., et al.: "Cystic kidneys: genetics, pathologic anatomy, clinical pictures, and prenatal diagnosis." Hum. Genet., 68:104-135, 1984.

ROBINOW SYNDROME

Synonymy

S. of distichiasis-lymphedema.

Syndromography

Clínic

This syndrome is characterized by two fundamental facts that give it its name: double row of eyelashes and lymphedema of the lower limbs. The double row of eyelashes can cause irritating eye problems, mainly keratitis. Lymphedema appears from the knees down and begins between the ages of 5-20, especially in adolescence.
There are reports of families that also present excess skin on the neck or pterygium colli.

Occasionally they may present ectropium of the lower eyelids, epicanthus, submucosal cleft palate, vertebral defects, congenital heart disease.

The prognosis is good, although there may be motor disability due to extradural cysts and leg deformity.

Paraclinical examinations

Radiology. In the spinal column, increased interpeduncular separation and the aforementioned defects are observed.

Lymphography. Dilated lymphatic vessels in both legs and absent, obstructed or deformed thoracic duct.

Syndromogenesis and etiology

The etiology is genetic with autosomal dominant inheritance proven by several family studies. It has been suggested that this syndrome is a rare form of bilateral hyperplasia.
There is an absence of meibomian glands, but the basic defect that explains both clinical effects is not known.

Literature

Kolin, T. et al.: *Hereditary lymphedema and distichiasis". Arch. Ophthal., 109: 980-981, 1991.

ROBINSON SYNDROME

Synonymy

S. tricho-dento-osseous. TDO syndrome.

Syndromography

Clinic

It is characterized by the patient having, from birth, curly hair. Craniofacial dysmorphism is limited to dolichocephaly, frontal bossing and square jaw. The teeth are small, separated, yellow due to enamel hypoplasia and increase in the pulp chamber (taurodontism), delayed bone eruption. Nails are exfoliating in 50% of cases.

Paraclinical examinations

Radiology. Slight or moderate increase in bone density, more evident in lateral views of the skull. Occasionally craniosynostosis.

Syndromogenesis and etiology

Of genetic etiology of autosomal dominant inheritance.
The basic defect is not known but apparently there are keratin and dentin defects. Currently, two forms have been proposed that differ in the partial or total presence and severity of clinical manifestations: TDO I and TDOI, which suggests the presence of at least two mutations or genetic heterogeneity.

Literature

Quattomanii, F. et al.: *Clinical heterogeneity in the tricho-dento- Ossious syndrome". Hum. Genet., 64:116-121, 1983.

ROTHMUND SYNDROME

Synonymy

S. of congenital poikiloderma.

Syndromography

Clínica

The clinical manifestations in these cases are mainly located in the skin and eyes. Patients have short stature in 54% of cases.

Skin alterations begin to appear between 3 months and one year of age, consisting of irregular erythema that progresses to linear telangiectasias, brownish pigmentation, depigmentation and punctate atrophy. They first appear at the level of the face and extend to the ears, buttocks and limbs until they acquire a marble appearance. They frequently have hypersensitivity to sunlight, mainly in early stages of life with the formation of blisters. Sometimes there is palmo-plantar hyperkeratosis with warty lesions.

50% have sparse hair or alopecia that includes eyebrows and eyelashes. The nails are atrophic or rough. The facies presents frontal bulging and depression of the nasal bridge. In the eyes, cataracts are the most common ocular finding (50% of cases) and develop rapidly; There may also be dystrophic changes of the conea. Occasionally microdontia and adontia, ectopic tooth eruption and caries, as well as mental retardation.

Paraclinical examinations

Diagnosis is clinical, since the histopathology of the lesions is not exclusive.

Syndromogenesis and etiology

The etiology is genetic with autosomal recessive inheritance criteria, although 70% of the cases studied are female.

The basic defect is unknown. The clinical variability in the reported cases suggests heterogeneity.

Literature

Hall, J.G.; R.A. Pagon and K.M. Wilson: “Rothmund-Thomson's syndrome with severe dwarfism.” Am. J. Dis. Child., 134:165- -169, 1980.

RUBINSTEIN-TAYBI SYNDROME

Synonymy

S. with thick thumbs.

Syndromography

Clinic

The syndrome can be recognized from birth due to the typical appearance, hands and feet, and short stature.

Patients present a craniofacial dysmorphism in which the following stand out: microcephaly, bulging or prominent forehead,

antimongoloid deviation of the palpebral fissures, epicanthus, hypertelorism, peaked nasal dorsum with nasal septum extending below the nasal wings, slight micrognathia, bushy eyebrows, long eyelashes, eyelid ptosis, nevus flammeus on the forehead. In addition, they have a strange grimace or smile, a high-arched palate and dental malocclusion. Strabismus, refractive errors, obstruction of the tear duct. Delayed closure of the fontanelle.

In the hands and feet the first finger is wide and in the hands they are angled, there may be brachydactyly of the fingers with clinodactyly and overlapping toes.

Other defects are: pectus excavatun, kyphoscoliosis and lordosis. Occasionally: congenital heart disease, supernumerary nipples, absence of corpus callosum. Mental retardation with IQ less than 50, motor and language delay.

Paraclinical examinations

Radiology. Increase of the foramen magnum and parietal foramina. Defects due to widening of phalanges.

Karyotype. To check the absence or presence of alterations tions.

Dermatoglyphs. Increase in thenar, interdigital and hypothenar patterns. There may be a summary groove.

Syndromogenesis and etiology

Although there is no doubt about its genetic etiology, the basic defect is unknown. Autosomal dominant inheritance has been suggested by family evidence. In some cases, de novo reciprocal translocation t(2,16) has been demonstrated.

Literature

Henneckan, R.C.M.; AC Stevens and J.J.P. Van de Kamp: "Etiology and recurrence risk in Rubinstein-Taybi's syndrome", Am. J. Med., Genet. Spp., 6:56-64, 1990.

RUSSELL-SILVER SYNDROME

Synonymy

S. of Russell's nanism. S. of intrauterine nanism- - craniofasciomandibular dysostosis.

Syndromography

Clínica

This syndrome is characterized by low stature that begins prenatally. Their growth pattern is parallel, but below the third percentile. Patients present asymmetry from birth, from the entire hemibody to limited to the skull or a limb or part of it.

There is skull (large)-face disproportion, which is triangular, frontal bulging and jaw hypoplasia, high-arched palate, crowded teeth, prominent eyes, bluish sclerae, long eyelashes, thin lips with downward corners, clinodactyly of the fifth finger, syndactyly of the second and third toes, café-au-lait spots, poor muscle development, hyperhidrosis, renal, ureteral and cardiac anomalies. They may present slight mental retardation.

Paraclinical examinations

Radiology. Delayed bone age, hip dislocation, shortening of the humerus, hypoplasia of the middle phalanx of the fifth finger, pseudoepiphysis at the base of the second metacarpal.

Gonadotropins in urine. Elevated in 10% of patients.

glycemia. Hypoglycemia after short periods of fasting in some cases.

Syndromogenesis and etiology

Of genetic etiology with heterogeneity. Karyotypes with 45.X mosaicisms have been published; 46,XY; XXY; trisomy 18 and 18p deletion.

Although the basic defect is unknown, it has been related to the finding in some cases of increased growth hormone with lack of response and short stature. Almost all cases are sporadic.

Literature

Escobar, V.; S. Gleiser and D.D. Weaver: "Phenotypic and genetic analysis of the Silver-Russell's syndrome." Clin. Genet., 13:278- -288, 1978.

SALDINO-NOONAN SYNDROME

Synonymy

S. of nanism, short ribs and polydactyly. S. of neonatal chondrodystrophy with polydactyly type II.

Syndromography

Clinic

At 20 weeks of gestation, manifestations can already be seen; It is a lethal form of neonatal chondrodystrophy.
Intrauterine nanism of short limbs and edema, with height less than 45 cm and weight less than 2.5 kg.; All cases have shortening of tubular bones, metaphyseal dysplasia with a splintered appearance, shortening of ribs and a narrow pelvis, with bone splinters; lung hypoplasia; congenital heart disease 88% of cases: transposition of the great vessels, coarctation of the aorta, septal defects, right hypoplasia, hypoplastic aortic arch. In 88% of cases, anal imperforation, intestinal malrotation, cystic fibrosis of the pancreas, lingual and epiglottic bifidity, situs inversus and esophageal atresia.

Genitourinary (88%): renal and cystic dysgenesis, hypoplastic and/or cystic ureters, small or absent urogenital orifice, septate uterus, hypoplastic double and cystic ovaries.

Paraclinical examinations

Radiology. The splintered appearance of the metaphyseal endings of the long bones and the pelvis and the hypermaturity of the epiphysis are almost pathognomonic.

Syndromogenesis and etiology

Genetics: autosomal recessive, with fetal lethality. Its frequency is around one case per 75,000 live births.

Literature

Saldino, R.M. and C.D. Noonan: “Severe thoracic dystrophy whit strikung micromelia, abnormal osseeous development, including the spine and multiple visceral abnormalities." Am.J. Roentgenol, 16:19, 1972.
Richardson, M.M.; TO THE. Beadet; M.L. Wagner et al. : "Prenatal diagnosis of recurrence of Saldino-Noonan dwarfism." J. Pediat. 91:467, 1977.

SANFILIPPO SYNDROME

Synonymy

S. of mucopolysaccharidosis II. S. of heparinuria. S. of polysaccharide oligophrenia. S. of oligophrenic polydystrophy.

Syndromography

Clinic

It starts between 1-3 years. Patients present severe mental retardation, which becomes very evident at school age.

Aggression, decreased attention, sleep disorders.

Later dementia, idiocy or imbecility. Minor somatic changes in the face. Tendency towards macroglossia, dry hypertrichosis on the head, poor mimicry. In some cases contracture is seen in the large joints and sometimes in the fingers. Good physical energy. Athetosis, seizures, autism.

Physical examination. Dwarfism (less severe than in other mucopolysaccharidoses). Moderate hepatosplenomegaly. Absence of corneal opacification. Transmission auditory disturbance with deafness. Coarse facies: depressed nasal root, coarse, abundant hair, triangular in section.

Delayed growth after 10 years.

Paraclinical examinations

Urine. Pathologically increased mucopolysacchariduria (2-9 times normal), formed in 70-90% by heparin sulfate.

Radiology. Similar bone abnormalities, but much less pronounced than in Hurler syndrome.

Medulogram. Metachromatic granulations in lymphocytes, plasma cells and reticulocytes.

Syndromogenesis and etiology

Mucopolysaccharide metabolism disorder. From the biochemical point of view, the enzymatic defect can be:
in type A heparinsulfamidase deficiency; in type B deficiency of N-acetyl-alpha-D-glucosaminase. There are two other types: type C due to acetyl CoA-alpha-glucosaminide-N-acetyl-transferase deficiency and type D due to N-acetylglucomi-na-6-sulfatesulfatase deficiency. These alterations lead to an excess of heparin and subsequent visceral overload of this substance.

The accumulation process is in lysosomes.

Of genetic etiology with non-allelic and hereditary heterogeneity.

autosomal recessive disease.

Literature

Sanfilippo, S.J. and R.A. Good: "Urinary acid mucopolysaccharides (AMP) in the Hurler syndrome and Morquio's disease." J. Pediat., 61:296-297, 1962.

Williams, H.E.: "Heritable disorders of mucopolysaccharide metabolism." Qual. Med. 106:306-311, 1967.

Bartsocas, C. et al.: "Sanfilippo type C disease: clinical findings in four patients with a new variant of mucopolysaccharidosis II."
Europ. J. Pediat., 130:251-258, 1979.

Kaplan, P. and L.S. Wolfe: "Sanfilippo syndrome type D."
Pediat., 140:267-271, 1987.

SCHWARTZ-JAMPEL SYNDROME

Synonymy

S. of myotonia-blepharophimosis. Myotonia with blepharophimosis and joint limitation. Blepharophimosis with generalized myopathy.

Osteochondromuscular dystrophy. S. of chondrodystrophy

myotonic.

Syndromography

Clínic

These patients present low height with postnatal onset, myotonia with fixed and sad facies, small jaw, muscular hypertrophy in half of the patients, and hyporeflexia. Blepharophimosis, with medial displacement of the external corners of the eye, long eyelashes inserted in two or several rows. Flattened nose. Articular limitation of the hips, wrists, fingers, toes and spine.

The hair set is low, ears set low, testicles small. pectus carinatum.

There may be mental deficiency and occasionally intrauterine growth retardation, delayed bone age, club feet, hip dislocation; microcornia and cataract.

During childhood they consult for orthopedic problems, decreased growth, and progressive myotonia with muscle loss.

Paraclinical examinations

Radiology. Platyspondyly, short neck, fragmentation and crushing of the femoral epiphysis.

Electromyogram. Compatible with myotonia.

Muscle biopsy, light and electron microscopy and histochemical examination. They show inconsistent muscle abnormalities.

Syndromogenesis and etiology

Of genetic etiology with autosomal recessive inheritance. The basic defect is unknown, although the fundamentally affected tissues are of mesodermal origin.

Literature

Fowler, W.M. et al.: "The Schwartz-Jampel's syndrome: its clinical, physiological and histological expression." J. Neurol. Sci., 22:127- -146, 1974.

SENIOR SYNDROME

Synonymy

Tubulointerstitial nephropathy and tapetum degeneration

retinal.

S. de Letterer-Senior-Loken.

Syndromography

Clinic

The first symptoms appear in the infant: thirst and polyuria. The fundus demonstrates progressive tapetum-retinal degeneration leading to blindness. Renal manifestations are characterized by nephronophthisis with minimal hematuria and proteinuria, and in the final stages during school age, hypertension and chronic renal failure are evident. There may be moderate to mild mental retardation and neurological symptoms such as cerebellar ataxia and deafness.

Paraclinical examinations

Urine metabolic tests. Proteinuria.

Cituria and other kidney function tests.

Electroretinography. Evidence of tapetum degeneration

retinal.

Radiology. Conical epiphysis.

Syndromogenesis and etiology

It is a syndrome of mitochondrial etiology. It is produced by the tubular absence of the enzyme cytochrome-C-oxidase of the respiratory chain, which is encoded by mitochondrial DNA. Deletion of this mitochondrial gene is also accompanied by encephalomyopathy.

In the pathological examination of the kidney, tubular atrophy and interstitial fibrosis are observed.

Mitochondria are dysmorphic with variation in size.

Molecular study has demonstrated mitochondrial DNA defects in kidney cells and leukocytes.

Literature

Bois, E. et P. Royer: "Assotiation the nephrophatie tubulo-instersticiells chronique et de degenerscense tapeto-retinienne: study genetics". Arch. Fr. Pediat., 27:471, 1970.
Senior. B.: A.J. Friedman and J.L. Braudo: “Juvenile familial nephropathy with tapetoretinal degeneration." Amer. J. Ophthal., 52:625, 1961.

SYNDROME 13 q

Synonymy

S. of partial monosomies of chromosome 13.

Syndromography

Clinic

Undetermined incidence. IM:1F sex ratio.

Severe microcephaly dysmorphology, Greek profile, wide and prominent nasal root, hypertelorism, epicanthus and eyelid ptosis. Short nasolabial distance, rabbit teeth. Small chin.
Large ears with deep helix canal. Hypoplasia of thumbs, abnormal implantation of toes. bi-scrotum
Fido.

Other malformations: microphthalmia, colobomas of the iris or retina, cataract. The most serious ocular anomaly is retinoblastoma.

Absence of the thumb, agenesis of the first metacarpal and fusion of the fourth and fifth, syndactyly. Heart disease, arrhinencephaly, aplasia of the falx cerebri, agenesis of the corpus callosum, periventricular atrophy, supernumerary ribs.

Neurology: they present severe encephalopathy and hypotrophy in height and weight. Mental retardation is very severe with IQ less than 50.

Prognosis: survival is difficult to assess. The severity of the clinical manifestations seems to correspond to the chromosomal deletion. Niebusr and Ottossen have defined three groups that, in summary, stand out for:

1. Craniofacial dysmorphology only.
2. Craniofacial dysmorphology and aplasia or hypoplasia of the thumb and ocular malformations.
3. Retinoblastoma in relation to the 13q21 deletion and relative rarity of the signs typical of groups I and II.

Paraclinical examinations

Karyotype0.

Kidney ultrasound.

Computerized axial tomography.

Bone survey.

Ophthalmological examination.

Dermatoglyphs. Anomalies related to defects in the hands.

Syndromogenesis and etiology

Chromosome imbalance due to deletion of segments of the long arm of chromosome 13. The ring of chromosome 13 is common.

In general, they are de novo defects.

Literature

Niebuhr, E. and J.Ottosen.: “Ring chromosome D(13) associated with multiple congenital malformations.” Ann. Génét., 16: 157- -166,1973.

18q- SYNDROME

Synonymy

S. of monosomy 18q2.

Syndromography

Clinic

The incidence has not been determined: more than 50 cases have been reported. Sexual relationship 2M:3E.

Dysmorphology: moderate microcephaly, depression of the middle third of the face with relative protrusion of the jaw. Carp-shaped mouth. Short nasolabial distance, poorly defined filter, everted lower lip. The pinna presents great development of the helix and antihelix, protruding antitragus, and atretic external auditory canal. There may be poor placement of the toes, long hands with protuberant fingertips.

Scrotal hypoplasia, small penis, labia atrophy
minors.

Other malformations: very common ocular malformations, amblyopia, optic atrophy, colobomas, rib anomalies, occult spina bifida. Cryptorchidism.

Neurology: variable mental retardation IQ less than 30, negative states may occur. A quarter of patients have IQs around 70. They present psychotic behavior and language difficulties. Seizures have been reported.

Prognosis: 10% of patients die in childhood, few survive to adulthood.

Paraclinical examinations

Karyotype.

Electroencephalogram.

Brain ultrasound.

Bone survey.

Eye exams.

Dermatoglyphs. Excess vortexes.

Immunological studies. One third of patients have IgA deficiency.

Audiometry.

Syndromogenesis and etiology

Chromosomal imbalance due to 18q deletion with breakpoint 21.2. 80% is a de novo deletion. 10% due to pericentric inversion or translocation in the parents. 10% are mosaicisms.

Literature

Grouchy, J. de: "The 18p-, 18q- and 18r syndromes." Birth Defects 5:74-87,1967.

XXXX SYNDROME

Synonymy

Tetrasomy X S.

Syndromography

Clinic

Dysmorphology: oval face with slight hypertelorism, bilateral epicanthus and mongoloid deviation of the palpebral fissures. There may be microcephaly. Clinodactyly of the fifth finger of the hands.

Other malformations: radioulnar synostosis in some cases.

Endocrinology: menstrual disorders.

Neurology: variable mental retardation.

Paraclinical examinations

Karyotype and sexual chromatin. Up to 3 Barr bodies per core.

Dermatoglyphs. Decrease in the total number of ridges.

Syndromogenesis and etiology

Chromosomal imbalance due to polyploidy of the X chromosome due to multiple nondisjunction.

Literature

Carr, D.H.; M.L. Barr and E.R. Plunkett: An XXXX sex chromosome complex in two mentally defective females". Canad.Med.Ass.J..84:132-137.1961.

Pena, S.D.J. et al: “A 48,XXXX female". J. Med. Genet., 11:211- -215, 1974.

XXXXX SYNDROME

Syndromography

Clinic

Dysmorphology: coarse-featured face with hypertelorism, epicanthus, mongoloid deviation of the palpebral fissures and marked flattening of the nasal root, resembling Down syndrome. Low set of ears. Short neck with low hair set at the nape of the neck. Narrow shoulders.
Clinodactyly of the little finger, abnormal implantation of the toes.

Other malformations: malformed teeth, congenital heart disease, patent ductus arteriosus, strabismus, limitation of flexion and supination of the elbow.

Neurology: variable mental retardation.

Paraclinical examinations

Radiology. Radioulnar synostosis, scoliosis, dislocation of hips.

Karyotype. Sexual chromatin (presence of up to 4 Barr bodies).

Radiology.

Echocardiography.

Dermatoglyphs. Decrease in the total number of ridges.

Syndromogenesis and etiology

Chromosomal imbalance due to polyploidy of the X chromosome due to various non-disjunction phenomena.

Literature

Berger, R. et al: "Syndrome 49,XXXXX". Ann.Pédiat.,20:965- -967. 1973.

XXXXY SYNDROME

Synonymy

S. of Fraccaro.

Syndromography

Clinic

Incidence: its frequency is little known.

Dysmorphology: oval face with significant hypertelorism and mongoloid deviation of the palpebral fissures with epicanthus, flattening of the nasal root, large mouth, large and protruding ears. Short neck with pterigum colli. Clinodactyly of the fifth finger of the hands. Small penis and scrotum.

Other malformations: strabismus, genital anomalies, ectopic testicles, limitation of elbow flexion and supination movements.

Neurology: profound oligophrenia in all cases (IQ between 20 and 50).

Paraclinical examinations

Karyotype.

Radiology.

Sexual chromatin. Presence of up to three bodies of Barr.

Radiology. Marked delay in bone age, coxa valga and the most frequent and important element is radioulnar synostosis or dislocation without synostosis.

Syndromogenesis and etiology

Imbalance due to polyploidy of the X chromosome due to double non-disjunction in oogenesis or spermatogenesis or simultaneous non-disjunction in both. Cases of mosaicism also show non-disjunction in the first divisions of the zygote.

Literature

Fraccaro, M. and J. Lindsten: “A child with 49 chromosomes". Lancet, II:1303.1960.

XXY SYNDROME

Synonymy

S. de Klinefelter.

Syndromography

Clinic

Incidence: 1.18 per 1,000 male newborns.

Dysmorphology: no significant dysmorphism. The tall stature is often referred to.

Malformations: testicular ectopia, hypospadias, hypoplasia of the penis and scrotum.

Endocrinology: gynecomastia around 12-13 years of age. Testicular atrophy with normal penile development. Before puberty, hormonal excretions are not modified. An increase in FSH is observed during puberty. Infertility. Decreased libido.

Neurology: intellectual development is normal. Mental retardation of varying degrees may correspond to the number of X chromosomes.

Paraclinical examinations

Sexual chromatin. There is presence of Barr bodies.

Dermatoglyphs. Increase in arc frequency (A).

Karyotype.

Syndromogenesis and etiology

Imbalance of X chromosomes due to aneuploidy as a consequence of meiotic non-disjunction or mitosis of the zygote. 80% of karyotypes are 47,XXXY, 20% 47,XXY/46,XY mosaicism. The 48,XXXY karyotype and mosaics related to it are accompanied by more severe hypogonadism and mental retardation is more frequent.

Literature

Boltshauser, E.; M. Meyer and T. Deonna: "Klinefelter syndrome and neurological disease." J. Neurol. 219:254-259,1978.

Ferrier, P.E.; S.A. Ferrier and G. Pescia: "The XXXY Klinefelter syndrome in childhood." Am. J. Dis. Child. 127:104-105,1974.

SYMPHALANGISM SYNDROME

Synonymy

S. of proximal symphalangism. Hereditary absence of proximal interphalangeal joints. Cushing's symphalangism.

Syndromography

Clinic

These patients present ankylosis of the proximal interphalangeal joints. Fusion of carpal and tarsal bones. In elbows the joint may be missing or only affected by radiohumeral synostosis.

The defect is accompanied by conductive deafness due to stapes synostosis.

Paraclinical examinations

Radiology. The bone alterations described are evident.

Audiometry. For the study of deafness.

Syndromogenesis and etiology

Of genetic etiology with autosomal dominant inheritance and variation in gene expressivity. The defects described seem to have a single explanation although the basic defect is not known.

Literature

Yesell. E.S.: "Symphalangism, strabismus and hearing loss in mother and daughter." New. Eng. J. Med., 263: 839-842, 1960.

SINOBRONCHITIS SYNDROME

Syndromography

Clinic

This syndrome is characterized by maxilloethmoidal sinusitis, bronchiectasis and dilation of the upper airways.

It begins between 3 and 7 years of age with recurrent lower respiratory infections. Bronchorrhea and occasionally hemoptysis, purulent rhinorrhea in the middle meatus, hypertrophy of the middle turbinate "bicolor" red above and pale-edematous below, sometimes there are polyps. It is accompanied by pondostural delay and sometimes lax skin and epidermal ear plugs.

Paraclinical examinations

Radiology. For the study of synopathy and bronchiectasis, which take the cystic form of glove fingers.

Bronchoscopy with fiberoptic bronchioscope0.

Electrolytes in sweat. Normal.

Cardiovascular study. Normal.

Syndromogenesis and etiology

It is a defect that appears to be congenital, with an unspecified etiology. The absence of elastic tissue in the trachea and main bronchi suggests a possibly monogenic common origin.

Literature

Legler, V. in Opitz-Schmid: Handbuch of the Kinderheikunde. Berlin, Springer, 1968, t. IX, p. 252.

Lemariey, A.: Oto-Rhino-laryngologie infantile. Ed. Masson, Paris, 1956, p.147.

Mounier-Kuhn, P.: "Le syndrome éthmoidoantrite et bronchiectasis' clinique: Etiologie. Hypotheses pathogenetiques." Ann. d'Otolaryngologie, 12:106, 1945.

Van de Calsyde, P.E: Linterdependance de lállergie intectee du nez et des bronches". Fortschr. Hais und Ohrenheik, 8:217, 1961.

SITUS INVERSUS SYNDROME

Synonymy

Laterality sequence.

Syndromography

Clinic

Often it is only a radiological finding, but in other cases dextrocardia accompanied by cardiac malformations can cause cyanosis, dyspnea on exertion, clubbing fingers, and endocardial murmurs. In these cases it may be accompanied by slow development. The liver is palpable on the left, as well as the ascending colon, cecum, and appendix. The stomach and duodenal arch on the right.

Paraclinical examinations

Radiology and ultrasound

Laparoscopy. It allows observing positional anomalies of the viscera.

Syndromogenesis and etiology

The primary defect is a failure of the normal asymmetry in morphogenesis, which results in both left

gives as right. The left laterality sequence is not accompanied by severe cardiac anomalies but is accompanied by polysplenia and has the same incidence in both sexes. The right-sided laterality sequence may be accompanied by asplenia, congenital heart and vascular disease, and situs inversus (Gremark syndrome).

It is two or three times more common in males than in females.

Literature

Cleveland, M.: *Situs inversus viscerum: anatomic study". Arch. Surg., 13:342, 1926.

Leiniger, C.R. and S. Gibbsson: "Transposition of viscera in siblings." J. Pediat., 37:195, 1950.

Smith, D.W.: Recognizable patterns of human malformation. 2 ed., Saunders, Philadelphia, 1976, p. 364.

SOTOS SYNDROME

Synonymy

Cerebral gigantism syndrome. S. of macrocephalic gigantism. S. of hypothalamic gigantism.

Syndromography

Clinic

Full-term children have a length of 55.2 cm and a weight of 3. 9 kg.

In the first 263 years growth is especially rapid, then it approaches the normal growth rate.

At 10 years old they have a size of about 14-15 years. From this age onwards, growth becomes normal, remaining, however, two standard deviations above the mean for chronological age.

Physical examination. Macrocephaly, dolichocephaly, prominent.

Antimongoloid palpebral fissures, hypertelorism. Prognathism with narrow jaw. Ogival palate. Premature eruption of teeth. Variable mental retardation and poor coordination.

Occasionally it can be accompanied by obesity, seizures and abnormal dermatological patterns. There are no currently demonstrable endocrinological alterations.

Paraclinical examinations

Karyotype. Normal (discard Fraya). See fragile X syndrome.

Radiology. Accelerated bone age.

Brain ultrasound. Ventricle dilation.

Computerized axial tomography. Dilation of the ventricular system.

Chromosome study. Normal.

Blood. Glucose tolerance test. abnormal in some cases.

Growth hormone levels in blood. Normal. Increase in 17-ketosteroids.

Syndromogenesis and etiology

It has been considered that this syndrome may be produced by factors that act in utero, by alterations in the function of the hypothalamic-pituitary axis or by lack of availability or autonomy of the somatotropic hormone. It is a genetic syndrome of possible autosomal dominant inheritance and it has been suggested that the gene is located in 3p21 or 6p21.

Literature

Schrander-Stumpel, C.T., J.P. Fryns and G.G. Hamers: "Sotos's syndrome and the novo balanced autosomal translocation t(3;6)(p21;p21)". Clin. Genet., 37: 226-229, 1990.

Sotos, J.F; PR. Dodge; D. Muirhead et al.: "Cerebral gigantism in childhood, a syndrome of excessively rapid growth with acromegalic features and a nonprogressive neurological disorder." New English J. Med., 271:109-116, 1964.

SPRANGER-WIEDEMANN SYNDROME

Synonymy

S. of congenital spondylo-epiphyseal dysplasia.

Syndromography

Clinic

It is a bone dysplasia of prenatal onset in which the child can reach a height between 32-52 inches. Facial dysmorphism shows flattened facies, molar hypoplasia.

Very short neck.

Barrel chest with pectus carinatum. Myopia with retinal detachment. Cleft palate. Muscle weakness, fatigue. Abdominal muscle hypoplasia. They have delays in walking, waddling gait. Decreased mobility of elbows, knees and hips.

Paraclinical examinations

Radiology (bone survey). AP and L vertebral column: flattened ovoid vertebral bodies with narrowing of the intervertebral spaces, odontoid hypoplasia, kyphoscoliosis, lumbar lordosis. Delayed mineralization of the epiphyses with a tendency to be flattened. Absence of mineralization of the centers of the pubis, talus, calcaneus and knees. Coxa stick.

Syndromogenesis and etiology

It is of genetic etiology with autosomal dominant inheritance.

The basic defect corresponds to the result of a mutation in the COL2A1 gene that affects type I collagen.

Literature

Anderson, I.J. et al. : "Spondyloepiphyseal congenital dysplasia: genetic linkage to type II collagen (COL2A1)". Am J. Hum. Genet. 46:896-901, 1990.

STICKLER SYNDROME

Synonymy

S. of hereditary arthro-ophthalmopathy.

Syndromography

Clinic

The clinical manifestations are very variable, presenting a dysmorphic facial pattern in which a round, asymmetrical face stands out with flattening of the structures of the middle part, epicanthus, sunken nasal root or saddle nose,

micrognathia. Ocular manifestations: congenital progressive myopia of up to 20 diopters, retinal detachment, vitreous and chorioretinal degeneration, cataract, glaucoma. Arthropathy corresponds to an increase in the size of the ankles, knees and wrists in the first years of life, joint pain and joint stiffness with rest, walking difficulties, premature and progressive arthritis, kyphosis and thoracic scoliosis, there may be joint hypermobility and hypotonia. Other malformations are: cleft palate in 15% of cases, 80% present sensorineural hearing loss.

Paraclinical examinations

Radiology. Moderate spondyloepiphyseal dysplasia. Bones with wide metaphysis and decreased width of the diaphyses. Coxa valga.

Audiometry. Deafness is checked.

Syndromogenesis and etiology

Of genetic etiology with autosomal dominant inheritance.

The basic defect appears to be determined by a mutation in the COL2A1 gene for type II collagen.

Literature

Francomano, C.A. et al.: The Stickler's syndrome: evidence for close linkage to the structural gene for type II collagen".
Genomics, 1:293-296, 1987.

SULFATIDOSIS SYNDROME

Synonymy

S. of sulfatidosis-mucopolysaccharidúria. S. of mucosulfatidosis. Lipidosis due to sulfatides. S. of metachromatic leukodystrophy. Scholz disease. Greenfield disease.

Syndromography

Clinic

It begins in the second year of life with psychomotor involution. Facies: normal without gargoyle, with "heavy" features. Growth: delayed psychophysical. Ichthyosis vulgaris.

Skeleton: oval vertebral bodies, coarse metacarpals, early closure of the longitudinal sutures of the skull.

Hepatosplenomegaly (50%). Eyes: tapetum-retinal degeneration and abolition of the electroretinogram (50%). Central nervous system: progressive mental deterioration, quadriplegia, nystagmus, brief seizures, deafness, blindness and progressive decerebration. Children usually die before the age of 4.

Three clinical forms are distinguished:

1. Infant form (type I). The most frequent. It manifests itself in the first or second year of life and begins with gait disorders, weakness of the limbs, which is replaced by hypertonia and hyperreflexia or flaccidity and areflexia, respectively indicative of pyramidal alteration or involvement of the anterior roots and peripheral nerves. It is accompanied by ataxia, nystagmus, tremor and seizures. Word and language disorders. Exceptionally, a cherry-red spot appears on the macula; more frequently optic atrophy or retinitis pigmentosa. Its course is subacute and death occurs in 2-3 years.

2. Juvenile form (type II). It begins at 4-10 years of age with ataxia and other neurological disorders that lead to death by the second decade of life.

3. Adult form (type III). It begins around 20-30 years of age in the form of psychosis or dementia. Life extends for decades.

Paraclinical examinations

Blood. Alder-Reilly anomaly in leukocytes (large neutrophilic granulations that stain much more intensely than normal granulations; they are purple or blue in color, which makes them different from eosinophils).

Urine. Absence of arylsulfatase and high proportion of mucopolysaccharides and metachromasia of the sediment.

Nerve biopsy. Metachromatic granules; nerve conduction speed, poor.

Electroencephalogram. Nonspecific alterations.

Electromyography and peripheral conduction velocity. Altered, they show alteration of the lower motor neuron.

Assessment of arylsulfatase A activity in white blood cells or fibroblasts.

Study of culture of fibroblasts from amniotic fluid. It is used to make prenatal diagnosis.

Cerebrospinal fluid. Hyperalbuminorrachia without alteration cellular rations.

Syndromogenesis and etiology

It is due to a deficiency of arylsulfatase A.B.C, which affects the metabolism of sphingolipids with excessive formation of cerebrosides (constituents of the myelin sheath) that cause degeneration of said sheath and extensive demyelination and destruction of oligodendroglia.

The metabolic blockade produces an abnormal storage of sulfate-cerebrosides in the white matter of the central and peripheral nervous system, as well as in some groups of neurons, liver, kidney, lung and wall of the gallbladder.

There is diffuse and symmetrical demyelination, with deposits of a material also being found in neurons and white matter that, in contact with toluidine blue or cresyl violet, metachromatically stains brown or reddish.

It is transmitted in an autosomal recessive manner.

The etiology is genetic.

Literature

Austin, J.H.: Mental retardation metachromatic leucodystrophy (sulfatide lipidosis) metachromatic leucoencephalopathy; in Medical aspects of mental retardation (Editor Carter). Ed. Ch.
C. Thomas, Springfield, 1965. p. 766.
Thieffry., A.; G. Lyon andP. Maroteaux: "Encéphalopathie métabolique associant une mucopolysaccharidose et une sulfatidose." French Arch. Pediat., 24:425, 1967.
Moser, H.W., in Stanbury-Wyngaarden-Frederickson: The Metabolic Basis of Inherited Disease. 3 Ed. Mac Graw-Hill, New York, 1972. p. 688.

TAYBI SYNDROME

Synonymy

S. oto-palato-digital type I.

Syndromography

Clínica

This syndrome is characterized by low height below 10th. percentile for your age. Slight mental deficiency. IQ of 75-90. Dysmorphism is characteristic especially in men due to thick and protruding eyebrows, supraorbital ridges

prominent, sloping ears and poorly modeled pavilions, wide and flattened nasal root, hypertelorism, mongoloid deviation of the palpebral fissures, increased mandibular angle, small mouth with downward commissures, cleft palate, facial anodontia and small tonsils, conduction deafness due to anomalies of the ossicles.

The trunk is small and the thorax has pectus excavatum. The hands have short, spatula-shaped thumbs. The feet have the same defect in the first gear. Limitation to elbow extension.

Paraclinical examinations

Radiology. Prominent frontal and occipital with thickening of the frontal bone and skull base with nasobasal angulation. Absence of frontal and sphenoid sinuses. Short, wide distal phalanges of the thumb and first joint with short third, fourth, and fifth metacarpals.

Audiometry. Study of deafness.

Syndromogenesis and etiology

Genetic etiology with semidominant inheritance linked to the X chromosome with variation in expressivity. There is research that supports the mapping of the OPDI gene to Xq28.

Literature

Biancalana, V. et al.: "Oto-palato-digital's syndrome type I: further evidence for assignment of the locus to Xq28". Hum. Genet., 88:228-230. 1991.

TAY-SACHS SYNDROME

Synonymy

S. of cerebro-macular degeneration. S. of lipoido-sis-ganglioside.

Syndromography

Clinic

It affects the Jewish population more frequently than other ethnic groups. The sexes are equally affected. It occurs between birth and 10 months (average 6 months) in apparently normal children. Insidious progression with feeding difficulties, weakness, growth retardation. Abnormal movements, seizures, hyperacusis, visual difficulties.

Physical examination. Hyperreflexia, spasticity in the limbs initially, later flaccidity and paralysis. Loss of vision.

In the fundus of the eye, red spots are seen in the macula.

Paraclinical examinations

Dehydrogenase and transaminase in serum and spinal fluid have abnormal values. Fructose-1-6-diphosphate is decreased in its specific activity.

Syndromogenesis and etiology

Hereditary metabolic defect of unknown nature, which causes the accumulation of a monosialoganglioside in proliferating glial cells.

Literature

Tay, W.: "Symmetrical changes in the region of the yellow spot in each eye of an infant." Trans. Ophth. Soc. U.K., 1:55-57, 1881.

Sachs, B.: "On arrested cerebral development, with special reference to its cortical pathology." J. Nerv. Ment. Dis., 14:541-553, 1887.

RUDIMENTARY TESTICLE SYNDROME

Synonymy

Dysgenic male pseudohermaphroditism. S. of Bergada.

Syndromography

Clinic

At birth the genitals correspond to a male with a small penis and scrotal bags with apparent cryptorchidism, the testicles are located in the abdomen or in the duct, but they are very small. In adults there is little development of secondary sexual characters and they are infertile. The existence of Mullerian derivatives is not proven.

Paraclinical examinations

Karyotype. 46, XY.

Syndromogenesis and etiology

This syndrome was described in 5 of 6 siblings born to consanguineous parents. If the etiology is genetic, it could be autosomal recessive.

Literature

Bergada, C. et al.: "Variants of embryonic testicular dysgenesis: bilateral anorchia and the syndrome of rudimentary testes. Acta Endocr., 40:521-536, 1962.

TYROSINEMIA SYNDROME

Synonymy

S. of hereditary tyrosinemia. S. of genuine tyrosinemia.

Syndromography

It was first described by Baber in 1956.

Clinic

It begins in the infant stage or before 4 years of age. It presents general manifestations such as anorexia, vomiting, lethargy, fever, abdominal distension, hepatosplenomegaly, jaundice. Diarrhea, melena, skin hyperpigmentation, signs of vitamin deficiency may appear; manifestations of liver cirrhosis with severe progressive failure that leads the patient to death; manifestations of renal failure with renotubular dystrophy and tyrosyluria (Baber), dehydration, potassium depletion and disorders of acid-base metabolism. It may be associated with the appearance of hepatomas.

Paraclinical examinations

Blood. Aminoaciduria and tyrosyluria. Plasma amino acids are usually elevated, particularly tyrosine and methionine, which can present figures of 5-10 times the normal value. Increased p-hydroxyphenylacetic acid. Glucosuria.

Syndromogenesis and etiology

It has not been convincingly demonstrated that all patients with tyrosinosis have the same disorder, although

They follow an autosomal recessive inheritance and in most cases present a deficiency of the enzyme parahydroxyphenylpyruvic oxidase.

Bibliography

Baber. M.D.: “A case of congenital cirrhosis of the liver with renal tubular defects, skin to those in the Fanconi syndrome."
Arch. Dis. Child., 31:335, 1956.

Lebental, E.: Gastroenterology and nutrition in Pediatrics. Barcelona, Ed. Salvat, 1985. p. 788.

NEONATAL TYROSINEMIA SYNDROME

Synonymy

S. of transient neonatal tyrosinemia.

Syndromography

Clinic

Newborns, and especially premature infants, if they ingest a large amount of protein (prepared with high concentrations of casein), eliminate tyrosine and p-hydroxyphenylpyruvic acid in the urine, which raises the suspicion of tyrosinemia, but this disorder is temporary and responds to protein restriction and the administration of vitamin C. There is controversy about the harmful disorders for the central nervous system of this entity, although the infant presents apathy.

Paraclinical examinations

Blood. High levels of tyrosine in plasma and urine.

Syndromogenesis and etiology

Lack of maturation of the enzyme p-hydroxyphenylpyruvic- -Oxidase. The importance of this syndrome is in relation to the differential diagnosis between this syndrome and hereditary tyrosinemia.

Treatment is with vitamin C.

The prognosis is excellent, 1metabolic defects disappear with treatment or spontaneously.

Literature

Avery, M.E.; C.L. Clow; J.H. Menkes et al.: "Transient tyrosinemia of the newborn: dietary and clinical aspects." Pediatrics, 39:378-384, 1967.

Lebenthal, E.: Gastroenterology and Nutrition in Pediatrics. Editorial Salvat, Barcelona, 1985. pp. 375; 789.

CONGENITAL TORTICOLIS SYNDROME

Syndromography

Clinic

It is due to shortening or contracture of the sternocleidomastoid muscle on one side of the body. It begins in the first month of life with tilting of the head and rotation of the occiput, and offers considerable resistance when trying to correct the position.

Exostosis can be palpated at the insertion point of the sternocleidomastoid on the clavicle, a downward inclination of the eyebrow on the affected side, a wide and vertically shortened face, and a deformed cranial vault.

Patients generally improve with muscle strengthening exercises, although sometimes surgical intervention is necessary (tenotomy of the muscle and its sheath).

Paraclinical examinations

Muscle biopsy. Vacualization and degeneration of the muscle fibers of the lower half of the sternocleidomastoid muscle can be observed in early stages. Later, connective tissue with normal muscle fibers remaining is observed.

Syndromogenesis and etiology

Several causes are responsible for this syndrome, there are different hypotheses: 1, intrauterine theory; 2, birth trauma theory 3, infectious theory; 4, neurogenic theory; 5, ischemic theory; 6, combination of the previous theories.

Treatment is through physiotherapy with good response to early treatment.

Literature

Adams, R.D.; D. Denny-Brown and C.M. Pearson: Diseases of the muscle. ed.2, Harper Row, New York, 1962.

Lidge, R.T.; R.C. Bechot and CN. Lambert: "Congenital muscular torticollis: etiology and pathology." J. Bone Joint Surg., 39A:
1165-1182, 1957.

Taylor, F: "Induration of sternomastoid muscle." Trans. Path.
Soc. London, 26:224-227, 1875.

TREACHER-COLLINS SYNDROME

Synonymy

Mandibulofacial dysostosis.

Syndromography

Clínic

From birth in these patients, the defects that characterize the syndrome are evident: malar and mandibular hypoplasia, antimongoloid deviation of the palpebral fissures, coloboma of the lower eyelid, malformation of the pinna with or without defects of the external canal and conductive deafness.

Occasionally there is cleft palate, incomplete soft palate and hair projection towards the cheek. Other occasional anomalies are: pharyngeal hypoplasia, upper eyelid coloboma, microphthalmia, macrostomia, microstomia, choanal atresia, absence of parotids, congenital heart disease, cryptorchidism.
Only 5% of reported cases present mental retardation. Auricular defects are highly variable from acrotia to blind fistulas or preauricular appendages that sometimes extend towards the tragus-oral line.

Paraclinical examinations

The diagnosis is eminently clinical.

Audiometry. For the diagnosis of deafness.

Syndromogenesis and etiology

Of genetic etiology with autosomal dominant inheritance and 100% gene penetrance; great variation in expressiveness. 60% are due to new mutations. In some cases balanced translocations and deletions have been reported. The gene has been located at 5q31-q34.

Literature

Dixon, M.J. et al.: "The gene for Treacher Collins's syndrome maps to the long arm of chromosome 5". Am. J. Hum. Genet., 49:17-22, 1991.

TRICHO-RHINO-EALANGIC SYNDROME

Synonymy

S. of Giedion. TRPS I.

Syndromography

Clinic

Patients present growth deficiency between the 3rd. and 10th. percentiles.

The facies is characteristic as that referred to in Langer-Giedion syndrome. The highlight is the pear-shaped nose, the filter is long and prominent, narrow palate, micrognathia, large and prominent ears. Small teeth with malocclusion and high frequency of cavities. Fine hair scattered and somewhat hypopigmented. Fine nails, fusiform fingers. Upper respiratory tract infections are common. Young adults can develop degenerative hip disease.

Paraclinical examinations

Karyotype. High ratio in search of 8q24 microdeletion.

Radiology. Flat coxa with occasional flattening of the head of the femur. Cone-shaped bone changes in the epiphyses in early childhood.

Syndromogenesis and etiology

There are three types of tricho-rhino-phalanx syndrome: one form with autosomal recessive inheritance and two autosomal dominant forms: type I and type II (Lauger-Giedion syndrome).

In type II, a deletion 8q 24.11-q 24.13 has been found. and in type I, 8q 24.12 deletion, which has allowed us to suggest that the 8qg 24.13 band is involved in exostosis.

The mental retardation that has been seen correlates with the chromosomal defect.

Literature

Hamers, A. et al.: "Severe mental retardation in a patient with tricho-rhino-phalangeal syndrome type I and 8q deletion." Europ. J. Pediat., 149:618-620, 1990.

TRICHORRHEXIS NODOSA SYNDROME WITH MENTAL RETARDATION

Synonymy

S. de Pollitt. S. of trichothiodystrophy-neuro-cutaneous.

Syndromography

Clinic

The patients described present apparent psychomotor retardation at 6 months, height and head circumference less than 3rd. percentile. On the face they have retrognathia and prominent ears.

The hair is curly, brittle, with knotty breaks. The eyebrows are thick, but the eyelashes are normal. Hypoplastic spoon-shaped nails. Neurological manifestations include eye jerks with head movements, spastic diplegia, absence of deep tendon reflexes. Sometimes there is photosensitivity, ichthyosiform changes with severe eczema in flexion folds.

Paraclinical examinations

Urine metabolic tests. Determine argininosuccinic acid in urine (argininosuccinic aciduria presents trichorrhexis nodosa; biotin deficiency).

Brain ultrasound or computed axial tomography. Cortical abnormality.

Determination of sulfur in hair. Reduction of its content (trichothiodystrophy).

Microscopic observation of hair. Pigment loss tation.

Blood determination of copper and ceruloplasmin. This test is performed on men. It allows it to be differentiated from Men Res syndrome.

Syndromogenesis and etiology

Autosomal recessive genetic etiology has been suggested and its basic defect could be related to the reduction of the protein sulfur content of the hair.

Literature

Pollit, R.J. and P.S. Stoiner: “Proteins of normal hair and of cystine-deficient hair from mentally retarded siblings.” Biochem. J., 122:433-444, 1971.

TRISOMY 13 SYNDROME

Synonymy.

S. of Patau.

Syndromography

Clinic

The incidence is 1 in 10,000 live births. Sex ratio: M:F, female is somewhat more common.

Dysmorphology: small skull, fontanelle and wide sutures. Bilateral microphthalmia, wide and flat nose. Low set ears. Hemangiomas on the face, forehead or neck, flexed fingers. Feet with protrusion of the calcaneus.

Other malformations: cleft lip, generally bilateral, with cleft palate. Postaxial polydactyly of hands and feet. Anophthalmia. Arynencephaly. Haloprosencephaly. Congenital heart disease. Polycystic kidneys, heterotopic islets of pancreas and spleen.

Neurology: seizures, hypotonia. They do not survive more than 130 days.

Paraclinical examinations

Karyotype.

Spine radiology. Vertebral anomalies, sacral hyperplasia and delayed bone age.

Blood. Hemoglobin electrophoresis: Gower-2 embryonic hemoglobin.

Syndromogenesis and etiology

There is a chromosomal imbalance. 80% due to free trisomy of chromosome 13 due to non-disjunction, 20% due to mosaicism, or due to translocation almost always t(13q 14q), generally de novo.

Literature

Patau, K. et al: "Multiple congenital anomaly caused by extra chromosome." Lancet, I:790,1960.

Schinzel, A.: “Autosomale Chromosomenaberationen”. Arch. Genet., 1979.

TRISOMY 18 SYNDROME

Synonymy

S. Edwards.

Syndromography

Clinic

The incidence is 1 in 8,000 live births. Sex ratio 4F:1M.
Prenatal history: postmature (420 g), poor fetal activity.

Dysmorphology: prominent occiput, microcephaly, faunistic ears, micrognathia. The hands are kept closed tightly, the index finger rides on the third finger and the fifth on the fourth. There is limitation to congenital abduction of the hips, protrusion of the calcaneus and ballated foot. First gear short and dorsiflexed. Short tőrax with excess skin. Defect of abdominal muscles, umbilical and inguinal hernia or dysostosis of the rectus abdominis.

Other malformations: cryptorchidism, clitoral hypertrophy, imperforate anus, congenital heart disease (septal defects and persistent ductus arteriosus). Abnormal lung segmentation. Meckel's diverticulum. Heterotopic islets of pancreatic tissue and spleen. Pyloric stenosis, omphalocele, intestinal malrotation and diaphragmatic hernia. Other malformations such as meningocele, radial defects, cleft lip are less common.

Neurology: severe growth retardation at birth.
There is hypotonia and then hypertonia. Severe mental retardation.
They survive for 2-10 months.

Paraclinical examinations

Karyotype.
Ultrasound.
Radiology. Narrowing of the pelvis and reduction of ossification points of the sternum are noted.

Syndromogenesis and etiology

80% have free trisomy 18, due to non-disjunction, with the risk increasing with maternal age; 10% are 46.XY(X)/47,XY(X),+18 mosaics and the other 10% are other types of structural or numerical aberrations involving chromosome 18.

Literature

Edwards, J.H. et al.: "A new trisomic syndrome." Lancet, I:787, 1960.

TRISOMY 9P SYNDROME

Synonymy

S. of doubling 9p.

Syndromography

Clinic

The incidence is estimated to be relatively high. Sex ratio 2F:1M.

Dysmorphology: microcephaly with brachycephaly. Anterior fontanelle wide and open to the metopic suture. Small, deep eyes, moderate hypertelorism. Characteristic nose, bulbous with prominent nasal septum. Short nasolabial distance, thin lips, downward lip corners with characteristic asymmetry when laughing or crying.

Concerned facies appearance. Short neck. Barrel chest. Characteristic hands with brachymesophalangy, clinodactyly of the fifth finger, proximal implantation of the thumb, dysplastic nails, hypoplastic especially on the feet. Genitals:
hypogonadism, delayed menarche.
Other malformations: cleft lip.
Neurology: delay in language development. IQ of 50.
The prognosis depends on the cytogenetic defect.

Paraclinical examinations

Karyotype0.
Radiology of hands, feet and pelvis. age delay
I mean.

Dermatoglyphs. Single palmar crease, brachymeso-phalangy, axial triradius in t", absence or fusion of triradii b and c, high frequency of thenar pattern.

Syndromogenesis and etiology

Excess arches. Chromosomal imbalance due to pure de novo trisomy 9p in half of the cases. Trisomy 9p associated with partial trisomy or monosomy of other chromosomes involved in balanced translocations, products of abnormal paternal segregations.

Literature

Lantigua, A. et al: "Trisomy 9p due to tandem duplication 9 (p12-p24). Karyotype phenotype correlation." Hum. Genet., 4:570- -577,1989.

Sutherland, G.R. et al: "Partial and complete trisomy 9. Delineation of a trisomy syndrome." Hum. Genet. 32:133-140,1976.

10Q PARTIAL TRISOMY SYNDROME

Synonymy

Duplication S. 10g2.

Syndromography

Clínic

Around 35 cases have been reported. Sex ratio 1M:1F.

Dysmorphology: microcephaly with brachycephaly. High and domed forehead. Round facies with flattened profile. Small palpebral fissures (bleparaphimosis), hypertelorism, epicanthus inverse, small nose with hypoplasia of the nasal bridge. Prominent lips, open mouth, high and arched palate. Small and retracted chin. Ears set low and rotated.
Short neck. Clinodactyly of the fifth finger. First hammer toe, separation between the first and second toes.

Other malformations: they are rare, heart disease and kidney malformations may occur. Palatine cleft.
Cryptorchidism.
Neurology: hypotonia, severe mental retardation. Delay of growth.

Prognosis: Death occurs in the first year in a third of patients, but seems to depend on the malformations that occur.

Paraclinical examinations

Karyotype.

Echocardiogram.

Kidney ultrasound.

Dermatoglyphs. Excess ulnar loops and flexion fold simple palmar

Syndromogenesis and etiology

Chromosomal imbalance due to trisomy 10q2 as a result of abnormal segregations from parents carrying balanced translocations involving this region of Chromosome 10.

Bibliography

Prieur, M. et al.: La trisomie 10g24-10qter". Ann. génét.. 18:217- -222, 1975.

Yunis, J.J. and O. Sánchez: "A new syndrome resulting from trisomy for the distal third of the long arm of chromosome 10". J. Pediat., 84:567-570,1974.

RADIAL ABSENCE-THROMBOPENIA SYNDROME

Synonymy

S. TAR.

Syndromography

Clínic

At birth, the absence or hypoplasia of the radius is evident, usually bilateral, which may be associated with ulnar hypoplasia and defects in the hands and lower limbs (legs and/or feet). It is curious that the thumbs are always present.

There may be congenital heart disease and other malformations such as kidney anomalies, spina bifida, Meckel's diverticulum. They are short patients with brachycephaly, nevus flameus on the forehead, strabismus, micrognathia, shoulder hypoplasia, and hip dislocation. 7% of patients may present mental retardation. Severe anemia in early childhood.

Paraclinical examinations

Hematological study. Thrombocytopenia, megakaryocytes absent in 66%, decreased in 12% and inactive in 12%. Leukemoid granulocytes in 62% of patients, especially due to hemorrhages. Eosinophilia in 53%.

Radiology. Dysostosis of bones of the limbs in relation to the congenital defect.

Ultrasound. To demonstrate other malformations genitas.

Syndromogenesis and etiology

Genetic etiology with autosomal recessive inheritance, the basic defect is unknown. The vascular defects in some cases suggest that the radial aplasia is primary and in others that the vascular defect determines the aplasia. Prenatal diagnosis is possible by observing bone defects in the upper limbs.

Literature

Hall, J.G: "Thrombocytopenia and absent radius (TAR syndrome)". J. Med. Genet., 24:79-83, 1987.

Luthy, D.A.; J.G. Hall and C.B. Graham: "Prenatal diagnosis of thrombocytopenia with absent radius" Clin. Genet., 15:495-499, 1979.

TURNER SYNDROME

Synonymy

Monosomy X S.

Syndromography

Clinic

The incidence is 0.4 per 1,000 live births.

Dysmorphology: small size from birth, may also present lymphedema of the hands and feet. The face is triangular, palpebral fissures facing downwards, epicanthus, blepharoptosis, lowered lip corners, poorly implanted teeth, hypoplasia of the jaw, low-set ears. Short neck with pterigum colli, low trident-shaped hair set. Shield thorax with teletelia. Cabitum valgus, pigmented nevi, hypoplastic nails. Infantile genitalia, incomplete development of secondary characters.

Other malformations: atrophy of the gonads, hypoplastic, bifid or reduced uterus in its cervical part. Congenital heart disease, especially coarctation of the aorta. Kidney malformations, rib anomalies. Myopia, cataracts, deafness.

Neurology: generally there is no mental retardation, although a higher frequency of these patients has been reported in institutions.

Paraclinical examinations

Karyotype and sexual chromatin. Generally absence of Barr bodies.

Dermatoglyphs. Increase in the total number of ridges among other characteristics.

Hormonal determinations in urine. Absence of estrogens and pregnandiol, increase in FSH greater than 40 US, low 17-ketosteroids.

Echocardiography.

Kidney ultrasound.

Bone survey. Kosowicz sign, ovalization of the carpus. Delayed bone age, small sella turcica.

Syndromogenesis and etiology

Imbalance due to aneuploidy of the X chromosome (karyotype 45, X) due to non-disjunction in gametogenesis or delayed anaphase in the first divisions of the zygote (mosaicism).

It can occur due to Xq isochromosome, rare cases of Xp isochromosomes or Xpó Xq deletions among other less frequent variants.

Literature

Chen, H.; D. Faigenbaum and H. Weiss: "Psychosocial aspects of patients with the Ullrich-Turner syndrome." J. Pediatr., 102:191,1981.

Turner, H.H. :"A syndrome of infantilism, congenital webbed neck, and cubitus valgus." Endocrinology, 23:566-574, 1938.

VAN BOGAERT-HOZAY SYNDROME

Synonymy

Non-mutilating acrosteolysis with facial dysmorphia.

Syndromography

Clinica

In these patients, facial dysmorphia includes flattening of the nasal root, thick cheeks, deformed ears, micrognathia, poorly implanted teeth, and absence of a beard. In addition, they present myopia and astigmatism and moderate to slight mental retardation.

The fingers and toes appear childish, there is a lack of development of the ulnar end.

Paraclinical examinations

Radiology. Acrosteolysis.

Syndromogenesis and etiology

Genetic disease with autosomal recessive inheritance. The basic defect is unknown.

VAN DER HOEVE SYNDROME

Synonymy

Dighton-Adair S. S. of osteogenesis imperfecta. S. of osteosatyrosis. S. of Löbstein. S. of the blue sclerae.

Syndromography

This syndrome manifests itself in a heterogeneous group of hereditary diseases that affect the connective tissue.

It is characterized by bone fragility that determines fractures that may be present from birth, causing skeletal deformities of variable severity. Lentinogenesis imperfecta, deafness, blue sclerae and soft tissue dysplasia.

Efforts have been made to classify these diseases into four groups based on the severity of the clinical and radiological manifestations, as well as the age of onset of symptoms, including genetic criteria. This classification has been quickly accepted by geneticists, but has been less popular among orthopedists.

Clinical manifestations:

Type I. Normal height, few deformities, blue sclerae, deafness in 50% of cases. Dentinogenesis imperfecta is rare and can give rise to a subtype.

Type II. Prenatal onset, very severe, lethal in the perinatal period, skull with very little mineralization, ring-shaped ribs, accordion femur, marked deformity of long bones, platyspondyly.

Type II. Progressive bone deformity not very marked at birth, blue sclerae at birth that lighten with age, dentinogenesis, deafness and short stature.

Type IV. Short stature, slight bone deformity, normal sclerae, dentinogenesis as a common feature and deafness in some cases.

Paraclinical examinations

Radiology. (bone survey). Translucent bones, thin diaphysis, deformities and consequences of accidents and fractures.

Blood. Calcium, phosphorus and alkaline phosphatase have normal values and this facilitates the diagnosis of bone metabolism defects.

Syndromogenesis and etiology

It is a connective tissue defect of genetic etiology due to mutations in the genes that encode type I collagen chains.

Type I. Autosomal dominant. Defect: decrease in type I procollagen.

Type II. Autosomal dominant and autosomal recessive. Defect: alterations in COL 11 and COL 1"2, substitution of glycine residues and small deletions in chain 2 (1).

Type II. Autosomal dominant. Defect: point mutation of the l (1) or 2 (1) chain. Also autosomal recessive, a mutation that produces pro 2 alterations (1).

Type IV. Autosomal dominant. Defect: alterations of 2 (1) due to point mutations, and small deletions.

Literature

Van der Hoeve, J. And A. De Kleyn: "Blaue Sclera, Knochenbruchigkeit und Schwerthorigkeit" . Arch. Ophth., 95:81-93, 1918.

Byers, P.H.: Disorders of collagen biosynthesis and structure. Chap. 134 in Scriver, C.R., A.L. Beaudet; W.S. Sly and D. Valle: The metabolic and molecular bases of inherited diseasaes.vol. III ed. 7 McGraw-Hill, Inc, 1995, pp 4039-4051.

McKusick, V.A.: Heritable Disorders of Connective Tissue. ed. 3, St. Louis, Mosby, 1966.

VAN DER WOUDE SYNDROME

Synonymy

S. cleft lip and/or palate-lip cysts. S. of labial dimples-hare lip.

Syndromography

Clinic

These patients present dimples on the lower lip in 80% of cases. Cleft lip cone without cleft palate, cleft palate and bifid uvula. Hypodontia, loss of central and lateral incisors, canines and/or bicuspids. Dimples can be fistulas of accessory salivary glands that can produce unpleasant saliva discharge for the patient, so it is recommended to remove them by surgery.

Paraclinical examinations

Eminently clinical diagnosis.

Syndromogenesis and etiology

Disease of genetic etiology with autosomal dominant inheritance with 80% penetrance.

Literature

Janken, P. et al.: The Van der Woude's syndrome in a large kindred: Variability, penetrance, genetic risks." Am. J. Med. Genet., 5:117- -123. 1980.

VATER SYNDROME

Synonymy

VATER Association.

Syndromography

Clinic

It is a sporadic polymalformative syndrome, composed of anomalies of: vertebrae, anus, trachea, esophagus and radius whose acronym gives the name VATER.

Vertebrae: malformed (70%), with increased number, fusions, hemivertebrae, generally presacral.

Anus: atretic (80%), or with malformations of the region. Tracheo-esophageal fistula (70%) and, occasionally, esophageal atresia.

Radius: dysplastic (65%) with polydactyly and/or lack or hypoplasia of the thumb; a lower limb is rarely affected.

Heart: congenital heart disease (53%), often due to interventricular defect. Navel: single umbilical artery (35%).

Other anomalies: rib and/or ear defects; large fontanelle: delay in prenatal development.

Paraclinical examinations

Radiology. In search of vertebral, radial axis and costal bone anomalies.

Abdominal ultrasound.

Echocardiogram.

Syndromogenesis and etiology

It would be caused by mesodermal damage before day 35 of gestation.

The etiology is genetic, sporadic with no known cause, possibly environmental. For Smith it does not constitute a syndrome but an " association"; On the other hand, Temtamy does call it a syndrome and thinks it may be genetic. Its frequency is one case per 75,000 live neonates. Around 35 cases have been published.

Literature

Quan, L. and D.W. Smith: The Vater association vertebral defect.
Anal atresia. Radial and Renal dysplasia. A spectrum of associated defects." J. Pediat. 82:104, 1973.
Say, B. and P.S. Gerald: “A new polydactyly, imperforate anus, vertebral anomalies syndrome." Lancet, 2:686, 1968.
Say, D. S. Balci; T. Pirnar et al.: "A new syndrome of dysmorphogenesis imperforated anus associated with polyoligodactyly and skeletal (mainly vertebral anomalies)." Paediat Act. Scand., 60;197, 1971.

Temtamy, S.A.; J.D. Miller: Extending the scope of the Vater association: Definition of a Vater syndrome." J. Pediat., 85:345, 1974.

WAARDENBURG SYNDROME

Synonymy

S. de Waardenburg-Klein. S. of Mende. S. of the first arch.

Syndromography

Clinic

It begins in childhood and is characterized by pigment disorders that include: white hair, partial albinism, poliosis. In the eyes, partial or total heterochromia of the iris, which sometimes shows a blue iris and another brown: outward displacement of the inner canthus, confluent eyebrows. Nasal root widened. Unilateral or bilateral deafness due to the absence of the organ of Corti.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. It is considered a genetic abnormality in the stapedial artery. It is part of a group of diseases that are the result of anomalies of the blood vessels that supply the first visceral arch in the embryo.

The gene has been located on the long arms of chromosome 2 (2q35). Three clinical types are recognized, denominated I, II and III, and seven allelic variants. The basic defect is related to molecular aspects of early embryogenesis.

Literature

De Hass, E.R.H. and K.E.W.P. Tank: "Waardenburg syndrome". Docum. Ophthal. (Den Haag) 21:239, 1966.
Waardenburg, P.: "Embryonic Fixation syndrome." A.J. Human Gen., 3:195, 1951.

WAGNER SYNDROME

Synonymy

Retinal hyaloid degeneration with palatoschisis.

Syndromography

Clínica

Affected children present facial dysmorphia: epicanthus, saddle nose, micrognathia and malformations such as apparent cleft palate or submucosa; defects of the hips and hyperextensible major joints. Ocular defects become evident around the age of 15: myopia, retinal hyaloid degeneration, vitreous degeneration and opacities of the posterior hyaloid membrane.

Paraclinical examinations

Those for the ophthalmological diagnosis of the syndrome drome.

Radiology. For the diagnosis of joint defects.

Syndromogenesis and etiology

Wagner syndrome does not appear to be caused by a COL2Al mutation and the existence of genetic heterogeneity between this syndrome and Stickler syndrome has been suggested.

It is of genetic etiology with autosomal dominant inheritance. nante.

Literature

Frandsen, E.: "Hereditary hyaloid-retinal degeneration (Wagner) in a Danish family." Acta Ophthalmol., 44:223-232, 1966.

WEAVER SYNDROME

Syndromography

Clinic

Children affected by this syndrome are large at birth and show accelerated growth that is evident in a minority of patients after a few months of age. They show slight hypertonia with progressive spasticity, hoarse crying and a dysmorphism in which a bifrontal diameter stands out.

large, flat occiput, ocular hypertelorism, epicanthus, antimongoloid deviation of the palpebral fissures, large ears, increased nasolabial distance and relative micrognathia, limb defects such as limitation in the extension of the elbow and knee joints, camptodactyly of the fingers with hypoplasia of the nails. Lax skin and fine hair. Umbilical hernia.

Paraclinical examinations

Radiology. Accelerated bone age. Distal width of the long bones.

Ultrasound and brain computed tomography. Due to abnormalities such as septumpellucidum cyst, cerebral atrophy and hypervascularization.

Syndromogenesis and etiology

Possible genetic etiology. There is a higher ratio of males than females. Cases with evidence of autosomal recessive inheritance have been reported.

Literature

Hall, B.D.: "Weaver's syndrome: expanded natural history." Prog.Clin. Biol. Res., 200:123-144, 1985.

WEISSMANN-NETTER SYNDROME

Synonymy

S. of dysplasia with curvature of the legs. Toxopa-diaphyseal chyosteosis. S. with curved legs with nanism.

Syndromography

Clínic

They are patients with low stature from prenatal onset, the intrauterine growth defect is accompanied by curvature of the femurs, although they also have saber tibias and slight curvature of the forearms. All of this is accompanied by a delay in starting to walk and sometimes mental retardation.

Paraclinical examinations

Radiology. Long bones curved and thickened, dural calcifications can be observed in the skull.

Syndromogenesis and etiology

It is a defect of genetic etiology of inheritance that is not well determined although with familial incidence. The basic defect is unknown.

Literature

Caffey, J.: Pediatric X-Ray Diagnosis. 5ed. Year Book, 1967, p. 815.
Krewer, B.: "Dismorphie jambiere de Weissmann-Netter (Toxo-pachy-osteose diaphysaire tibioperonienne) chez deux vrais jumueaux." Presse Med., 69:419, 1961.
Weissmann-Netter, R. et L. Stuhl: “D'une osteopathie congenitale eventuellement familiale". Presse Med., 62:1618, 1954.

WERNER SYNDROME

Syndromography

Clinic

This is a syndrome that is characterized by premature aging with a life expectancy of 47 years. They are short individuals with a slow growth rate in childhood.
They have loss of subcutaneous fat with patches of hardened skin particularly on the face and limbs, skin ulcerations and atrophy of distal limbs. There is thickening of the subcutaneous fibrous tissue with thin dermis.

muscle hypoplasia. Osteoporosis and arteriosclerosis with calcification.

The hair turns gray and there is premature baldness around the age of 20 and cataracts at the age of 25 with retinal degeneration.

Premature loss of teeth. Hypogonadism and decreased fertility. Voice changes secondary to vocal cord atrophy. Liver atrophy. Organic brain syndrome with sclerosis.

Metastatic calcifications. 44% develop adult-onset diabetes and there is increased excretion of hyaluronic acid.

Paraclinical examinations

Skin biopsy. The changes already described are observed.

Karyotype. Chromosomal breaks and rearrangements.

Syndromogenesis and etiology

This syndrome has a genetic etiology of autosomal recessive inheritance. The basic defect is related to a decrease in the activity of the enzyme 5-hydroxymethyluracid-glycosylase, chromosomal instability, a decrease in the rate of cell division and premature aging not related to age.

Literature

Demple, B. and L. Marrison: "Repair of oxidative damage to DNA: Enzymology and Biology". Anu. Rev. Biochem., 63:915-948, 1994.
Salk. D.: "Werner's syndrome: A review of recent research with an analysis of connective tissue metabolism, growth control of cultured cells, and chromosomal aberrations." Hum. Genet., 62: 1-5, 1982.

ZELLWEGER SYNDROME

Synonymy

S. cerebro-hepato-renal.

Syndromography

Clinic

Most affected children are born in a pelvic position and make little progress. They may develop jaundice and bloody stools.
The fundamental clinical signs are: severe hypotonia, respiratory difficulties, difficulty sucking, seizures, hyperammonemia, hepatomegaly with cirrhotic changes, renal cysts mainly of the glomerulus.

The defects are mainly located in the brain:

macrogyria and polymicrogyria, incomplete myelination, gross defects of early brain development. Death occurs between the first day and six months of birth.

Physical examination. They present characteristic craniofacial dysmorphism: flattened occiput, brachycephaly, high forehead, small orbits, flattened facies, epicanthus, micrognathia, redundant skin on the nape of the neck.

Persistent ductus arteriosus, septal defect.

Paraclinical examinations

Metabolic studies of:

Pipecolic acid: increased in plasma and urine.

Phytanic acid: increase in serum and urine.

Very long chain fatty acids: increase in plasma and urine.

Increase in bile acids.

Deficiency of plasmalogens in serum and fibroblasts.

To the electron microscope. Absence or reduction to less than 10% of peroxisomes in hepatocytes and fibroblasts.

Syndromogenesis and etiology

It presents autosomal recessive genetic etiology. It is a metabolic defect that is characterized by the absence or decrease in the number of peroxisomes. The finding of microdeletion involves 7q 11.12-qll.13, which has suggested the location of the gene in this region in a second case with a pericentric inversion (7)(p12q11. 13), confirmation of this assignment is suggested.

The genetic defect seems to be related to the passage of peroxisomal enzymes through the peroxisome membrane, so long-chain fatty acids are not metabolized.

Literature

Kelly, R.I. : "Review: The cerebrohepatorenal syndrome of Zellweger. Morphologic and metabolic aspects." Am. J. Med.
Genet., 16:503, 1983.Naritomi, K. et al.: "Gene assignment of Zellweger's syndrome to 7ql1.23: report of the second case associated with a pericentric inversion of chromosome 7." Hum.
Genet., 84:79-80, 1989.
Wanders, R.J.A. et al.: "The inborn errors of peroxisomal beta-oxidation: a review." J. Inherit. Metab. Dis., 13:4-36, 1990.

ZINSSER-ENGMANM-COLE SYNDROME

Synonymy

Cole-Raush-Toomey S. Dyskeratosis congenita with hyperpigmentation.

Syndromography

Clínic

It is a syndrome described only in men. It begins at school age and progresses until puberty. The alterations occur in the skin and mucous membranes: dark gray reticular hyperpigmentation in the neck and genitals, leukoplakias in the mouth, anus and penis.
The nails have striations and partial anonychia. Palmoplantar hyperhidrosis and thin, light-colored sparse hair.

These patients suffer from chronic blepharitis and epiphora due to lacrimal obstruction.

Other clinical manifestations are: dental anomalies, hypogonadism, esophageal stenosis with dysphagia, osteoporosis, thrombopenia, aplastic anemia, and mental retardation.

Paraclinical examinations

Skin biopsy. Atrophy and pigment-laden macrophages.

Hematological study. Pancytopenia.

Syndromogenesis and etiology

Of genetic etiology. The gene has been assigned to Xq28 by linkage analysis with DNA markers as well, so X-linked recessive inheritance is proposed.

Literature

Connor, J.M. and R.H. Jeague: “Dyskeratosis congenita: report of a large kindred." Brit. J. Derm., 105:321-325, 1981.

RELATED PEDIATRIC SYNDROMES

Syndromes with Pterygium

Antecubital pterygium.
Multiple pterygium.
Lethal multiple pterygium.
Popliteal pterygium.
Pterygium colli isolated.
Pterygium X-linked.

ALICE IN WONDERLAND SYNDROME

Synonymy

S. of depersonalization. S. of unreality.

Syndromography

Clinic

It is characterized by depersonalization, body image disorders and double personality, alteration in the sense of the passage of time and space. Disorders in the sensory sphere with minimization. Metamorphosis.

Syndromogenesis and etiology

It is observed in certain cases during a migraine attack, in epilepsy, poisoning and drug addiction, schizophrenia, and those who take hallucinogenic drugs. It is also observed during febrile delirium (probably due to alterations in cerebral circulation), and tumors or lesions of the temporal lobe.

Literature

Carroll, L.: Alice's adventures in Wonderland. New York, 1865.
Todd, J.: "The syndrome of Alice in the Wonderland." Dog. Med. Ass. J., 73:701-704, 1955.

ALZHEIMER SYNDROME

Synonymy

S. of presenile dementia. Alzheimer's dementia.

Syndromography

Clínica

It predominates in women and begins in the fifth to sixth decades of life. It is characterized by gradual loss of anterograde memory; At first the patient is aware of this alteration and worries. Later in the course of the syndrome, the patient is disheveled in his clothing, disoriented in time and space, has a need for sleep and sometimes short periods of euphoria. Lack of attention, speech disorders and inactivity. Occasionally hemiparesis, tremor and sometimes seizures.

Physical examination. Some alterations of the central nervous system, frequently hyperreflexia or tendon hyporeflexia.
Non-intense hypertonia.

Paraclinical examinations

Encephalogram. Internal hydrocephalus.

Syndromogenesis and etiology

Apparently dominant hereditary condition with highly variable expressiveness. Alterations have been discovered in three genes from different chromosomes, which apparently produce an abnormal protein and could be related to the pathophysiology of this syndrome.

The etiology is unknown.

Literature

Alzheimer, A.: Uber Einen Eigengartigen Schweren Krankheits: Prozess der Hirnrinde". ZBL., Nervekh, 25:1134, 1906; 30:177-179, 1907.
Gordon, E.B.: “Serial electroencephalogram studies in presenile dementia.” Brit. J. Psychiat., 114:779-780, 1968.
Crapper, D.R.; S.S. Krishnan and S. Quittak: "Alluminium, neurofibrillary degeneration and Alzheimer's disease." Brain, 99:67-68, 1976.

ANOREXIA NERVOSA SYNDROME

Synonymy

S. of nervous apepsia.

Syndromography

Clinic

It is observed in young women, especially in adolescence or early adulthood. They are previously healthy patients who, for fear of gaining weight, do not want to eat food.
In addition, they have disorders in their ingestion and aversion to food. As they lose weight, they maintain anorexia and say they "feel fat", although in reality they are emaciated. The population at risk is made up of young white women from the middle or upper class; it is rarely observed in poor, black or oriental people.

The patients deny being hungry, fatigued or thin and are often physically active and are frequently in exercise programs that they complete like a ritual. Occasionally they may have vomiting and abdominal symptoms: feeling of fullness, belching, pain, nausea, constipation and sometimes diarrhea. Amenorrhea and disorders in temperature sensation with intolerance to cold. It is accompanied by depression and other neurotic disorders.

Paraclinical examinations

Blood. Anemia and leukopenia. Hypokalemia and hypoalbuminemia.

Dosage of pituitary and ovarian hormones.

Syndromogenesis and etiology

Although the cause is unknown, psychological factors, personality disorders, problems in family relationships and interpersonal communication between family members are considered.

Abnormalities in the concentration of neurotransmitters in the cerebrospinal fluid have been reported,0 but it is not known whether these changes are primary or secondary.

It is possible that the occupation is some predisposing element since it is more common in dancers, as well as in runners.

Literature

Schwabe, A.D.. et al.: "Anorexia nervosa." Ann. Intern. Med.. 94:371, 1981.

ANXIETY-TENSION SYNDROME

Synonymy

S. of anxiety-tension-fatigue.

Syndromography

Clinic

This syndrome is manifested by tension in the voice and anxious gaze. The patient sits at the end of the chair and rests his sweaty palms to dry them on it. Somaic manifestations that are given by: muscle hypertension, headache, dizziness, blurred vision, feeling of a lump in the throat.
It is also accompanied by cardiac distress, spastic colon, diarrhea or constipation, flushing of the face or other psychological symptoms.

In their physical appearance they look good, they are intelligent, perfectionists, they are successful in their social and economic life, despite which they are not satisfied.

Syndromogenesis and etiology

It is considered that they suffer from a "chronic alarm reaction" with internal changes. Because there is an obstacle in their sensitivity or inner feeling.

Literature

Freedman, A.M., and H.I. Kaplan: Comprehensive Textbook of Psychiatry. Baltimore, Williams & Wilkins, 1967.

AKINETIC SYNDROME

Synonymy

S. hypokinetic.

Syndromography

Clínic

It is manifested by muscle rigidity associated with a slight alteration of voluntary movements. The speech is slow and monotonous, there may be tremors in the upper limbs, preferably hands and forearms, few movements. Disorders in standing and walking. The facies has a fixed expression with some eye winks.

Physical examination. Cogwheel sign, normal reflexes, absence of Babinski.

Syndromogenesis and etiology

It is considered a clinical form of paralysis agitans (Parkinson's syndrome) .

Literature

Grinker, R.R., and A.L. Sahs: Neurology. ed., 6, Springfield, Thomas, 1966.

CAPGRAS SYNDROME

Synonymy

S. double personality.

Syndromography

Clínic

It is more common in women. Patients adopt another personality and are convinced that the new one has replaced the original one and behave as such (if they are married the spouse always accepts it) and in this way they reject any explanation that attempts to rule out this fact.

Syndromogenesis and etiology

It is a psychopathological complex associated with paranoid psychosis or affective schizophrenia. There may be other psychiatric conditions. No organic alteration could be demonstrated, which is why it is considered a functional disorder. It can be classified as an identification agnosia.
The etiology is unknown.

Literature

Capgras, J., and J. Reboul-Lachaux: "Illusion des sosies dans un delire systematise chronique." Bull. Soc. Clin. of Med. Ment..,. 11:6. 1923.
Enoch, M.0., W.H. Trethowan, and J.C. Barker: Some Uncommon Psychiatric Syndromes. Baltimore, Williams & Wilkins, 1967.

CLERAMBAULT SYNDROME

Synonymy

S. of erotomania.

Syndromography

Clinic

It generally occurs in healthy women who mistakenly think that a man is deeply in love with them. The chosen person is a prominent figure, older than the patient and her husband, and whom they only know superficially. Other disorders that coexist are: interpretations and reactions of acceptance or rejection. In this type of love, the patient secretly keeps her feelings from her friends and husband. They frequently attempt a sexual relationship with the person of their fantasy or affection.

Syndromogenesis and etiology

Erotomania can exist as an autonomous entity or be a premonitory syndrome of other psychoses and sometimes be a symptom of paranoid psychosis. It is considered a psychiatric disorder that is due to dissatisfaction associated with a feeling of rebellion.

Literature

De Clerambault, G.G.: "Les psychoses passionelles, Oevre psychiatrique." Paris, Presses Universitaires, 1942.
Enoch, M.O., W.H. Trethowan, and J.C. Barker: Some Uncommon Psychiatric Syndromes. Baltimore, Williams & Wilkins, 1967.

COTARD SYNDROME

Synonymy

S. of delusion of denial".

Syndromography

Clínic

Patients complain that everything is negative: their possessions, their physical or intellectual vigor, different organs (heart, intestine, blood) or the entire body. They also think that the world does not exist for them and paradoxically they feel that they are immortal. Other megalomelancholic ideas may be present.

Syndromogenesis and etiology

This syndrome is observed especially in patients with manic-depressive conditions, but also in certain syndromes of the nervous system.

Literature

Cotard, J.: Du délire des negations. Arch de Neurol., 4:152-282, 1882.

Freedman, A.M., and Kaplan, H.I.: Comprehensive Tex Book of Psychiatry. Baltimore, Williams & Wilkins, 1967.

COUVADE SYNDROME

Syndromnography

Clínic

It is seen in men whose wives are pregnant.

Symptoms generally begin around the third month of the wife's pregnancy and are preferably characterized by gastrointestinal symptoms: loss of appetite, nausea, vomiting, toothache, morning sickness, constipation or diarrhea.

They can be accompanied by anxiety. Intestinal inflammation is rare and is only seen in Simpson syndrome. Unlike cases of disappointment at not being pregnant, patients with Couvade syndrome never have the idea that they are pregnant.

Syndromogenesis and etiology

This neurotic disorder is the equivalent of the Couvade ritual that many primitive peoples have practiced since ancient times. This ritual consists of the father, when the wife is in labor, imitates the pains and receives the same attention as her and after the birth they lie in bed with the child to receive the congratulations of their neighbors and friends.

The etiology is unknown.

Literature

Enoch, M.O.; W.H. Trethowan and J.C. Barker: Some Uncommon Psychiatric Syndromes. Baltimore, Williams & Wilkins, 1967.

SENILE DEMENTIA SYNDROME

Syndromography

Clinic

It is observed in both sexes and begins after age 60. The onset is insidious and is manifested by loss of recent memory and a marked interest in the past. The power of concentration decreases and there are changes in behavior, paranoid tendencies and depression. Loss or disinterest in personal appearance, occasionally: hallucinations and delusions (see chronic brain syndrome). There may be focal symptoms.

Physical examination. Decreased reflexes, lazy pupils, loss of sensitivity to the tuning fork.

Paraclinical examinations

Electroencephalography.

Pneumoencephalogram.

Arteriography

Computed axial tomography and/or nuclear magnetic resonance.

Leukocyte culture and chromosome count. Loss of chromosomal material is seen.

Syndromogenesis and etiology

It is considered to be a different genetic process than arteriosclerotic dementia. It is transmitted by autosomal dominant inheritance with low penetrance. Decreased chromosomes have been found, and in women alterations of the X chromosome. The basic defect is unknown.

Literature

Larsson, T.. T. Sjögren, and G. Jacobson: *Senile dementia, a clinical sociomedical and genetic study". Acta Psychiat. Scand. (suppl. 167), 39:1-259, 1963.

Nielsen, J.: “Chromosomes in senile dementia.” Brit. J. Psychiat. 114:303-309, 1968.

GANSER SYNDROME

Synonymy

S. of nonsense. S. pseudodemential. S. of apparent idiocy. Acute hallucinatory mania.

Syndromography

Clinic

The symptoms appear linked to different circumstances that seem to precipitate them, but sometimes the patient reports that he does not know what this relationship could have been, and in turn mixes that response with other nonsensical ones. They show apathy, indifference, lethargy, semi-stupor and disorientation. Sometimes there are somatic conversion symptoms manifested by motor and sensory disorders, and problems may appear in movements, tone (flaccidity or contracture), headache, back pain, areas of anesthesia. Visual or auditory hallucinations. Frequently disorientation, childishness.

Syndromogenesis and etiology

It is possible that this is a dissociative hysterical reaction.

This syndrome must be differentiated from people who feign illness, from true dementia or pseudodementia and from some variants of schizophrenia. It has been observed in prisoners of war.

The etiology is unknown.

Literature

Ganser, S.J.: “Euber einen eigenartigen hysterischen Dammerzustund.” Arch. Psychiat. Nervlkrank., 30:633-640, 1898.

Enoch, M.D., W.H. Trethwan, and J.C. Barker: Some Uncommon Psychiatric Syndromes. Baltimore, Williams & Wilkins, 1967.

HPERSOMNIA SYNDROME

Synonymy

S. de Gelinaud.

Syndromography

Clínica

It gradually begins with an irresistible desire to sleep and the patient wants to stay in bed all day and sleeps for many hours, days or weeks (not associated with catalepsy, hypnagogic hallucinations or narcolepsy). Difficulty waking up, irritable, dizzy and somewhat mentally slow afterwards.

Concomitant manifestations: obesity, mild diabetes insipidus, diffuse hyperhidrosis, salivary secretion disorders, acromegaly, hypogenitalism.

Paraclinical examinations

Blood. Polycythemia, paradoxical reaction to adrenaline, hypoglycemia.

Syndromogenesis and etiology

Multiple conditions can cause this syndrome: brain tumors, destructive or inflammatory lesions of the brain stem, menarche or the menstrual cycle, underlying psychogenic alterations, hypothalamic dysfunction.

Kleine-Levin syndrome can be considered a particular form of hypersomnia and Pickwickian and Elpenor syndromes should also be considered, although for other mechanisms.

The basic lesion seems to be in a center located in the thalamus, hypothalamus or midbrain, which when injured causes diencephalic dysregulation.

Literature

Bonkalo, A.: "Hypersomnia, a discussion of psychiatric implication based on three cases." Brit. J. Psychiatr., 114:69-75, 1968.

POST-PARTUM BLOODS SYNDROME

Synonymy

S. of the third day. S. of postpartum depression. S. of postpartum crying.

Syndromography

It is manifested by short-term crying attacks without any reason or precipitating factor and not associated with depressive feelings. They generally occur on the third day after delivery and end on the tenth day. It is accompanied by fatigue, increased irritability, sleep disorders, insomnia, the feeling of considering the husband a stranger and, on the contrary, excessive care with the baby.

Syndromogenesis and etiology

Stressful or endocrine factors have been mentioned.

The etiology is unknown.

Bibliography

Savage, G.: "Observations on the insanity of pregnancy and childbirth." Guy's Hosp. Rep., 20:83-117, 1875.
Yalom, I.D., D.T. Lunde, R.H. Moos, et al.: “Post-partum blues Arch. Gen. syndrome, a description and related variables" Psychiat., 18:16-17, 1968.

MÜNCHAUSEN SYNDROME

Synonymy

S. of hospital dependency.

Syndromography

Clinic

It predominates in men in a 3:1 ratio and the average age is 39 years. Patients pretend to have severe urgent illnesses, serious and serious underlying problems, and sometimes report gruesome or aggressive symptoms for their body, and, however, they maintain a behavior that avoids medical advice.

They are admitted to multiple hospitals and produce complex clinical pictures, undergoing expensive and often painful examinations and even multiple surgical interventions.
They wander around different hospitals and not only consume a large amount of medical resources but apparently feel satisfied with the concern they cause to the specialists.

This phenomenon has created so many problems that doctors at different congresses or meetings have identified the same subjects and have made the decision to create a "blacklist" with the generals and photos of this type of patients to prevent them from repeating different or similar stories and causing them to waste so much time, with some results but not enough.

The name Münchausen syndrome comes from Baron Katrl Friederick von Münchausen (11720-1797), who spent his life making people believe that he had carried out countless acts of military heroism. even inflicting wounds on themselves to make their imaginary stories more convincing.

Syndromogenesis and etiology

Patients are victims of fear or anxiety and that is why they deceive the doctor. This deception, according to research, occurs because when the specialist asks them if they suffer from stomach pain, they, afraid of being diagnosed with some serious illness, think that an affirmative answer (the truth) directs the doctor towards that serious illness that they fear so much, and consequently, they answer that the pain is in the back.

The syndrome seems to be produced by different psychopathological entities, different from self-mutilation and those who feign symptoms or alterations that they do not have, but it also includes these conditions, as well as antisocial personalities, neuroses, brain damage or other conditions.

The etiology is unknown.

Literature

Asher, R.: "Münchausen's syndrome." Lancet, 1:339-341, 1951.
Ireland, P., J.D. Sapira, and B. Templeton: "Münchausen's syndrome, review and report of an additional case." Amer. J.Med. 43:579-592, 1967.
Spiro, H.R.: “Chronic factitious illness, Münchausen's syndrome.” Arch. Gen. Psychiatr., 18:569-579, 1968.

OTHELLO SYNDROME

Synonymy

S. of sexual jealousy. S. of morbid jealousy. S. of psychotic jealousy.

Syndromography

Clínic

It can affect both sexes, with a predominance in men, generally in the fourth decade of life. The patient begins to have a feeling of infidelity on the part of his spouse, whom he accuses of small events or a particular episode that, according to him, proves the events that occurred previously. He meticulously and obsessively searches for evidence and continually subjects his partner to interrogations and pressures her to confess her guilt. His sexual activity increases and if he encounters any rejection he interprets it as proof of infidelity; He has great tension and depression that causes him problems at work. He frequently has outbursts of violence and explosions, particularly against his partner.

Syndromogenesis and etiology

This syndrome is considered a special variety of paranoia, but it may be a feature of manic-depressive psychosis.

It has also been observed in alcoholics and epileptics.

Literature

Todd, J., and K. Dewhurst: "Othello syndrome; a study in psychopathology of sexual jealousy" J. Nerv. Ment. Dis., 122:367-374, 1955.

Enoch, M.O., W.H. Trethowan, and J. C. Barker: Some Uncommon Psychiatric syndromes. Baltimore, Williams & Wilkins, 1967.

SLEEP PARALYSIS SYNDROME

Syndromography

Clinic

Symptoms appear at the moment of beginning sleep or upon waking up, and are manifested by a state of anxiety accompanied by hypnagogic hallucinations. The person is unable to speak or scream and only emits a moan that appears in the middle of the dream and when he wakes up he has a sensation of darkness, aware of his surroundings. The attacks pass in 1-2 minutes, but subjectively this sensation remains for a while, which can be interrupted by a sudden movement or because someone touches the patient. This syndrome has some differences from night dread syndrome.

Syndromogenesis and etiology

It is considered to be due to a deep psychological disorder.

The etiology is unknown.

Literature

Mitchell, S.W.: *On some of the disorders of sleep." Virginia Med. Monthly, 2:769-781. (Feb.), 1876.

Schneck, J.M.; H. Fuseli: "Nightmare, and sleep paralysis." J.A.M.A.. 207:725, 1969.

PSYCHOGENOUS POLYDIPSIA SYNDROME

Synonymy

S. of compulsive ingestion of water.

Syndromography

Clínic

More common in women (80%), it can begin at ages between 6-60 years, but generally between 35-60 years. It is a common trait in children with a history of walking while sleeping.

These patients do not have a stable and satisfying sexual life and complain of dramatic organic situations that require consulting a doctor.

The onset can be vague or sudden, frequently associated with menopause, illness, stress of various types. The patient ingests excessive amounts of water for hours or days. Vasopressin does not affect polydipsia. There may be periods of remission for months; polyuria, they are generally obese and sometimes there is fluctuation in blood pressure.

Paraclinical examinations

Plasma. Marked decrease in osmolality.

Urine. The ability to concentrate urine varies considerably and is decreased in many patients.

Syndromogenesis and etiology

It is considered a psychotic compulsion associated with other minor psychotic manifestations, which have small or severe neurotic behaviors.

Literature

Bralow, E.D., and H.E. by Wardener: "Compulsive water drinking." Quat. J. Med., 28:235-258, 1959.

Goldberg, M.: “Abnormalities in the renal excretion of water." Med. Clin. N. Amer., 47:915-933. 1963.

PSYCHOGENIC PURPLE SYNDROME

Synonymy

S. of hysterical purple. S. of hysterical hemorrhage. S. of psychosomatic hemorrhage.

Syndromography

Clinic

It occurs in patients with major emotional disorders and/or psychotic manifestations; more frequently in women. The symptoms consist of hemorrhages or bruises related to certain complex psychic experiences. Injuries are produced to imitate the body of Christ, Muhammad or that of other religions. Sometimes spontaneous hemorrhages in healed wounds associated with strong emotional experiences, when remembering those experiences. They would be equivalent to menstruation in various parts of the body (nose, armpit) or in the hands.

Paraclinical examinations

Blood. Coagulation studies to rule out other types of purpura.

Psychoanalysis. It can reproduce the symptoms if the psychotic condition that produces it can be stimulated.

Syndromogenesis and etiology

Opinions are divided about its pathogenesis:
supranatural origin, hysterical conversion, self-induced phenomenon.

The etiology is unknown.

Literature

Hyde, J.H.: “A contribution to the study of bleeding stigmata.” J. Cutan. Dis., 15:557, 1897.
Agle, D.P., and 0.D. Ratnoff: "Purpura as a psychosomatic entity." Arch. Int. Med., 109:685-694, 1962.

SIMPSON SYNDROME

Synonymy

S. of hysterical distention of the abdomen.

Syndromography

Clinic

Exceptional in men (see Couvade syndrome), it occurs in women and is manifested by abnormal distension of the pseudopregnant abdomen.

Physical examination. Depression of the diaphragm and lordosis of the spine are found in many cases.

Paraclinical examinations

Anesthesia. Under its effect the clinical picture disappears, but it returns as soon as they wake up.

Syndromogenesis and etiology

It is considered a manifestation of disappointment at not being pregnant, as well as other psychotic symptoms.

The etiology is unknown.

Bibliography

Simpson, J.: Clinical Lectures on Diseases of Women. Edinburgh, Black, 1872, p. 363.
Enock, M.0., W.H. Trethowan, and J.C. Barker: Some uncommon psychiatric syndromes. Baltimore, Williams & Wilkins, 1967.

ALCOHOL WITHDRAWAL SYNDROME

Syndromography

Clinic

It is observed in chronic alcoholic individuals when they stop drinking alcohol; Symptoms are more severe in patients who have been drinking for a longer period of time.

It is manifested by tremors or agitation, intense anxiety, insomnia, lack of concentration, feeling of unreality. In severe cases: trunk ataxia, nausea, vomiting, diarrhea, abdominal cramps. 25% of patients suffer from hallucinations and incoherence in communication. Isolated seizures or grand mal may be the first symptoms in combination with severe tremors and hallucinations.

Physical examination. Signs of dehydration, tachycardia, pupillary dilation, sweating, fever, increased blood pressure.

Paraclinical examinations

Liver tests.

Blood. Serum pyruvates and pyruvate tolerance curve.

Syndromogenesis and etiology

The withdrawal of alcohol is the fundamental cause and may be associated with a deficiency of the vitamin B complex or other nutritional deficiencies caused by the previous state.

Literature

Josserand, A.: "Le syndrome gros coeur et emphysame important complications viscerale de lálcoholisme et facteur de surmortalite masculine." Lyon Medical, 189:42-44, 1953.

TRANSSEXUALISM SYNDROME

Synonymy

S. of transsexual psychopathy.

Syndromography

Clinic

It predominates in men and the desire for sexual transformation through surgical techniques and hormones is around 3-7 times more frequent, which manage to change the patient's appearance and achieve complete identification with the other sex.
In the case of men this involves: castration, penectomy, and surgical construction of an artificial vagina. In women mastectomy, hysterectomy. In both adequate hormonal treatment.

Syndromogenesis and etiology

There are no organic or genetic alterations, although there are patients who show some sexual anomalies. Psychological and social factors have been considered at an early age, in games combined with some biological factor. In other cases, it starts from paranoid states.

Literature

Friedreich, J.: “Versuch Einer Literargeschichte der Pathologic und Therapie der psychischen Krankheiten.” Würzburg, 1930.
Pauly, I.B.: “Male psychosexual inversion: Transsexualism: a review of 100 cases.” Arch. Gen. Psychiat., 13:172-181, 1965.

"DRESSING DISORDER SYNDROME"

Synonymy

S. of fashion.

Syndromography

Clinic

Described by Frankerburg and Yurguelun-Todd in 1984, it has been described in different countries and seems to be increasing rapidly, according to different reports. It always occurs in women and is characterized by the increasingly accentuated dependence on acquiring absolutely unnecessary clothing and clothing accessories, to the point that some of the patients give away very expensive clothing shortly afterward despite it having been economically difficult for them to acquire them. One percent of the risk is found among young people aged 17-25, who show true adoration for elite brands and extraordinarily expensive items without caring about their usefulness or quality. Paradoxically, this group is the one with the fewest resources

economic and therefore seven out of ten are considered victims of the so-called hidden poverty", a new situation that consists of the practical impossibility of obtaining employment in accordance with their technical capacity.

This syndrome is usually associated with other nervous imbalances; However, it often appears as the only sporadic or chronic clinical form linked to depressive states. In its most acute phase, it turns those affected into automatons with consumerist stimuli and very low levels of security and self-esteem. The concern for the external image enters the field of obsessional neurosis, interrelating with the feelings of insecurity and lack of self-esteem already mentioned.

Syndromogenesis and etiology

The disproportionate cult of image tries to respond to a strong feeling of internal dissatisfaction, transferring low personal self-esteem to an purchasing potential far above what one's economy allows. This gap ultimately translates into a marked deterioration and, frequently, the breakdown of family relationships.

The “fashion syndrome" thus becomes, along with alcoholism and drug addiction, an important and dangerous cause of imbalance.

WITTMAAK-EKBOM SYNDROME

Synonymy

S. of Ekbom. S. of paresthetic crural asthenia. S. of restless legs. S. of irritable legs.

Syndromography

Clínic

It is manifested by recurrent attacks of strange sensations in the legs as if they were dragging or crawling and sometimes also in the thighs and feet, and sensations within the muscles or bones that prevent the patient from maintaining the affected limb in a fixed position. It can be a single member or bilateral with predominance in one of them. It rarely occurs in the upper limbs or hands and if it does, the sensation is slight. True pain is rare. It can occur in short periods or hours. The unpleasant sensation disappears with movement, only to reappear when the patient returns to bed, which is often an important cause of insomnia.

Physical examination. Coldness is only found in the feet and legs.

Syndromogenesis and etiology

It seems due to psychological factors, since the symptoms frequently appear, especially during boring film, television or theater programs. Iron deficiency anemia or a certain lack of irrigation should be evaluated.

It has been considered autosomal dominant inheritance.

In pregnant women, the syndrome occurs in 10% and disappears after childbirth.

Literature

Willis, T.: The London Practice of Physick. London, Bassett Crooke, 1685, p. 404.

Wittmaak, T.: Pathologie und Therapie der Sensibilitat-Neurosen.
Leizip, Schafer, 1861.

Ekbom, K.A.: "Asthenia crurum paraesthetica (*irritable legs"); a new syndrome consisting of weakness, sensation of cold and nocturnal paresthesia in legs, responding to certain extent to treatment with priscol and doryl". Note on paresthesia in general. Acta. Med. Scandinav., 118:197-209, 1944.

RELATED PSYCHIATRIC SYNDROMES

Syndromes of the burlesque or fantastic character

Ahasvero.

Munchausen.

Giobbe.

Gianno.

Leprechaunism.

Nocturnal polyphagous.

Cherubism.

Pickwickian.

Alice in Wonderland.

From the iceberg.

Of albatrosses.

LEOPARD.

Rapunzel.

Oblomowism.

Undine.

Of greed.

VATER.

PLUM ABDOMEN SYNDROME

Syndromography

Clinic

This complete syndrome only exists in men. It is characterized by a special wrinkling of the abdominal wall that gives the syndrome its rather striking name.

It is observed in neonates and this aspect may provide the key to early diagnosis. This is due to the presence of intestinal loops under the skin. Many cases reach adolescence or the beginning of adult life. There is bilateral cryptorchidism. When it occurs in females, this symptom is naturally missing.

Paraclinical examinations

Radiology. It shows a marked dilation of the bladder with an apical diverticulum that sometimes reaches the umbilicus and appears to be fixed to it. The ureters are greatly dilated, but the degree of hydronephrosis often appears disproportionately small.

Syndromogenesis and etiology

This rare anomaly is characterized by the absence of lower abdominal midline muscles, urinary tract dilation, and bilateral cryptorchidism. However, it has not been possible to demonstrate that it is due to sex-linked inheritance. The etiology is unknown.

ALBRIGHT-BUTTLER-BLOOMBERG SYNDROME

Synonymy

S. of primary hyperchloremic acidosis. S. de Albright- - Buttler. S. of primary tubular acidosis. S. of rickets- vitamin D-resistant.

Syndromography

Clinic

It is a process in which a sustained metabolic acidosis of renal origin, disproportionate to glomerular insufficiency or in its absence, is the initial pathophysiological event. The term secondary renal tubular acidosis refers to the production of the same defect by a specific kidney disease or by other pathological processes.

In young people it is characterized by: rickets, with growth retardation resistant to usual doses of vitamin D, muscle hypotonia and paralysis due to hypokalemia. Hyperplasia of the dental enamel with early deterioration of the teeth.

In adults, osteomalacia, nephrolithiasis and nephrocalcinosis predominate, leading to kidney failure.

Paraclinical examinations

Urine. Renal acidosis, hypercalciuria, ammoniagenesis deficiency due to distal tubule failure. Hyperphosphaturia.

Blood. Hyperchloremia. Hypophosphatemia. Frequent increase in alkaline phosphatase.

Syndromogenesis and etiology

The fundamental lesion is renal tubular failure.

There are two types: type I, which is transmitted by autosomal dominant inheritance, and type II, which is transmitted by autosomal recessive inheritance. The recessive form is probably due to a defect in carbonic anhydrase B. In the dominant form

There is an increase in the permeability of the distal tubule to hydrogen, which causes an increase in the back diffusion of this anion into the blood, which gives rise to acidosis. This alteration is responsible for hypercalciuria, hypercaliuria, nephrocalcinosis and renal failure.

Literature

Rodríguez-Soriano. J. and C.M. Edelmann: "Renal tubular acidosis", Ann. Rev. Med., 20, 363. 1969.

Albright, E., A.M. Buttler, E. Bloomberg: “Rickets resistant to vit D therapy.” Amer. J. Dis. Child., 54:529, 1937.

ALPORT SYNDROME

Synonymy

S. otooculorenal. S. of familial hemorrhagic nephritis. S. of hematuria-nephropathy-hereditary deafness. Dickinson S.

Syndromography

Clínic

This syndrome is characterized by: 1, hematuric nephropathy, either glomerulonephritis or pyelonephritis; 2, central type deafness; 3, congenital cataracts. Spherophakia and lenticone.

The disease begins at an early age and hematuria predominates. Kidney failure progresses slowly and usually ends in its final phase in the second or third decades of life.

Ophthalmological complications are spherophakia and lenticone; There are also thrombocytopathies, hyperprolinemia and brain dysfunction. The intensity of deafness does not seem to be related to kidney failure and can appear several years after birth. This is neurogenic deafness, due to damage to the auditory nerve or the organ of Corti itself.

Physical examination. In women, hematuria as a single manifestation. Deafness of neurogenic origin. Kidney failure during pregnancy. In men, hematuria, albuminuria, cylindruria, recurrent pyelonephritis, generally deafness of neurogenic origin, due to alteration of the auditory nerve or the organ of Corti. Occasionally congenital anomalies in the eyes (cataracts, spheropakias).

Paraclinical examinations

Urine. Proteinuria, cylindruria, hematuria. Sometimes renal glycosuria and aminoaciduria (serine, alanine, taurine, glycine, proline) .

Blood. Hyperprolinemia. Sometimes leukopenia with relative lymphocytosis. Increase in 2-globulin.

Hearing tests. They demonstrate neurogenic deafness.

Syndromogenesis and etiology

The syndrome is transmitted by autosomal dominant inheritance with variable expressivity for the different pleiotropic manifestations of the gene.

It is more common in males, but more serious in females.

Substitution of serine for cysteine has been demonstrated in the alpha-5 chain of type IV collagen, which correlates with the abnormalities of the syndrome.

The basic defect has not been fully defined.

Literature

Alport, A.C.: "Hereditary familial congenital hemorrhagic nephritis." Brit. Med. J., 1: 504-506, 1927.

Spear, G.S.: "Hereditary nephritis (Alport's syndrome), 1983." Clin. Nephro., 21:3, 1983.

Barker. OF. et al: Identification of mutations in the COLAAS collagen gene in Alport syndrome. Science, 248:1224-1227, 1990.

Dickinson, W.H.: Diseases of the kidney and urinary disorders. Part 2:376, London, Longmans, 1875.

Dockhorn, R.J.: "Hereditary nephropathy without deafness." Amer.J. Dis. Child., 114:135-138, 1967.

CRUSH SYNDROME

Synonymy

Crush-syndrome. S. of Bywaters. S. myorenal. S. of muscle destruction. S. compression. S. of ischemic muscle necrosis.

Syndromography

Clínica

After a violent accident with crushing of muscle fibers, acute renal failure with anuria, hypertension and uremia (traumatic uremia) occurs.

State of shock. Necrosis and parenchymal lesions of the liver (which translate into jaundice) and kidney.

Pain at the site of trauma, thirst, nausea, oliguria, anuria and shock.

Physical examination. There is damage to the affected areas. Hyper-blood pressure. Moderate to severe signs of shock.

Phlogistic swelling is seen in the traumatized area.

Paraclinical examinations

Blood. Anemia, leukocytosis, hyperkalemia, uremia, creatininemia. Decrease in sodium.

Urine. Alkaline pH, proteinuria, creatinuria, hemoglobinuria and myoglobinuria. Proteinuria, glucosuria, granular casts.

Electrocardiogram. Signs of hyperkalemia.

Syndromogenesis and etiology

Crush syndrome is caused by extensive disintegration of striated muscles (trauma, electrical flare, ischemic muscle necrosis).

This injury causes prolonged tissue ischemia.

The clinical manifestations are presented by the release of large amounts of myoglobin and necrosis of the kidney and liver parenchyma accompanied by shock.

Literature

Bywaters, E. and D.D. Beal: "Crush injury with impairment of renal function" Brit. Med. J., 1:427-432, 1941.

Hackredt-Boret's Laboratory Reports, Germany. 1917 quoted by: Paxson, N.E, Golub, L.J., and Hunter R.M.: The crush syndrome in obstetrics and gynecology. J.A.M.A., 131:500-04, 1946.

BARTTER SYNDROME

Synonymy

S. of chronic familial hypokalemia. S. of juxtaglomerular hyperplasia-hyperaldosteronism-hypokalemic alkalosis.

S. of primary hyperprostaglandinism.

Syndromography

Clínica

This syndrome begins in the infant, with symptoms of polyuria, polydipsia, anorexia, vomiting, constipation, accompanied by muscle hypotonia, sadness and symptoms of malnutrition.

Blood pressure is normal, growth is slow in the first years.

Delay in bone maturation is also observed and sometimes there is tetany; Intermittent paralysis with general adynamia and diffuse pain sometimes accompanied by headache, vertigo and paresthesia. Nocturnal polyuria.

The clinical picture is reminiscent of hyperaldosteronism but without hypertension. Other conditions that are accompanied by chronic metabolic hypokalemia and Fanconi syndrome, infantile renal acidosis and hypokalemic nephritis should be excluded.

Patients can die from dehydration, uremia or added bacterial diarrhea. Bartter syndrome has also been reported in adolescents and young adults of both

sexes.

Paraclinical examinations

Blood. Hypokalemia (1.5-2.5 mEq/L) and hypochloremia. Hyponatremia, metabolic alkalosis. Renin and angiotensin II levels are elevated in plasma.

Urine. It only shows a concentration deficit and sometimes proteinuria. Hyperaldosteronuria.

Kidney biopsy. It is normal, but hypertrophy of the juxtaglomerular apparatus is

Syndromogenesis and etiology

It is transmitted by autosomal recessive or sporadic inheritance. Its production mechanism is doubtful: there seems to be a lack of vascular response to angiotensin, with consecutive hyperplasia of the juxtaglomerular apparatus and hyperaldosteronism. As is known, hypokalemia stimulates the excessive production of PGE2, which has a vasodepressant action. This elevation, as well as that of bradykinin, explain the normality of blood pressure. The increase in PGE is considered the primary defect and is due to the increase in the synthesis of this substance by the interstitial cells of the kidney. Prostaglandin or its precursors induce vasodilation and stimulation of the renin-angiotensin-aldosterone system, which in turn causes the suppression of the K-Na-ATPase enzyme, and consequently inhibits the tubular reabsorption of sodium in the proximal renal tubuli.

Literature

Bartter, F.C. et al.: Hyperplasia of the juxtaglomerular complex hyperaldosteronism and hypokalemic alkalosis. A new syndrome". Am. J. Med., 33, 1962.
Royer, P., et al.: Pediatric Nephrology (Spanish trans.). Ed. Toray, Barcelona, 1975, p. 40.
Simopoulos, A.P. yF.C. Bartter: "Growth characteristics and factors influencing growth in Bartter's syndrome." J. Pediat., 81:56. 1972.

BLADDER NECK SYNDROME IN WOMEN

Synonymy

S. of contracture of the neck of the bladder. See S. of prostatic obstruction in women.

Syndromography

Clínica

This syndrome is characterized by difficulty initiating urination, followed by a thin stream and a terminal drip. Urinary infections are frequently associated.

It presents a greatly increased residual bladder volume, with bladder enlargement and diverticulum formation.

Neurological examination allows us to exclude neurogenic bladder.

Paraclinical examinations

Cystography and cystourethrogram. They allow the residual volume of urine to be determined, which is very important.

Cystoendoscopy. Allows calibration of the bladder neck.

Syndromogenesis and etiology

The syndrome is due to a congenital narrowing of the bladder neck and a dysfunction of the sphincter muscles of the bladder neck. Among the acquired causes, surgical interventions on the bladder neck and chronic neck infections are considered.

It must be differentiated from stenosis caused by extrinsic compression (pelvic tumors, pregnancy, etc.).

Literature

Ward, J.N., R.W. Lavengood, and J.W. Draper: "Pseudo bladder neck syndrome in women." J. Urol., 99;65-68, 1968.

EPSTEIN SYNDROME

Synonymy

Idiopathic nephrotic S. Lipoid nephrosis. Membranous glomerulonephrosis.

Syndromography

Clinic

This syndrome is generally observed in children or young people, more frequently in males, between 1 and 6 years old, but it can occur at any age and is characterized by anorexia, vomiting, diarrhea and dyspnea.

Physical examination. Edema, which takes over the eyes and spreads, becoming a true anasarca, pallor, sometimes hydrothorax that can be bilateral, and ascites. In some cases high blood pressure.

Paraclinical examinations

Blood. Marked hypoalbuminemia with alpha 2 and beta hyperglobulinemia, hyperfibrinogenemia, hyperlipidemia, elevated urea, and a flat glucose tolerance curve. Accelerated erythrocyte sedimentation.

Urine. Marked albuminuria, hematuria, hyaline and granular casts.

Basal metabolism. Low.

Kidney biopsy. Thinning of the basement membrane, proliferation of endothelial cells. If the lesion progresses it can cause hyalinization and fibrosis.

Syndromogenesis and etiology

In adults it has been seen after streptococcal glomerulonephritis. In children there seems to be a fa-
miliary.

It has been considered a syndrome produced by an autoimmune mechanism.

Pathogenically, an initial alteration of capillary permeability in the glomerulus occurs, which is responsible for massive albuminuria. This implies hypoalbuminemia and, as a consequence, generalized edema due to the drop in colloid osmotic pressure in the plasma with all its clinical consequences.

unique

The etiology is unknown.

Literature

Epstein, A.A.: "Concerning the causation of edema in chronic parenchymatous nephritis; methods for its alleviation." Amer. J. Med. Sci., 154:638-647, 1917.
Drummond, K.N.; A.F. Michael; R.A. Good, et al: "The nephrotic syndrome of childhood; immunologic, clinical and pathological correlations." J. Clin. Invest., 45:620-630. 1966.

EPIDEMIC HEMORRHAGIC FEVER SYNDROME

Synonymy

Korean hemorrhagic fever S. S. of nephrosishemorrhagic nephritis.

Syndromography

Clinic

This syndrome has been reported in Manchuria, Korea, Siberia, eastern Russia, Czechoslovakia and Hungary, with two annual peaks related to some seasons, but in general it is endemic. It occurs in all ages and both sexes.

The onset is abrupt and is characterized by: headache, fever, chills, anorexia, nausea, vomiting and back pain.

Physical examination. First day: conjunctival injection, redness of the skin, especially on the face and neck. Second day: Petechiae appear on the face, oral mucosa, conjunctiva and axillary regions. Third day: periorbital edema. Fourth day: the fever disappears, but hypotension and oliguria are found. From the tenth day onwards: diuretic phase that lasts days or weeks with fluctuations between shock and hypertensive crises with pulmonary edema in relation to fluid balance. Convalescence lasts 3-12 weeks and recovery is gradual.

Paraclinical examinations

Blood. Elevated hematocrit; leukocytosis above 50,000 with immature leukocytes. Thrombocytopenia. After-

hydroelectrolyte balance lathes. During the oliguric phase, elevation of urea and creatinine.

Urine. Proteinuria.

Syndromogenesis and etiology

It is considered due to a viral infection transmitted by the bite of arachnids similar to the scabies spider.

The etiology is unknown.

Literature

Dennis, L.H., and M.E. Conrad: “Accelerated intravascular coagulation in a patient with Korean hemorrhagic fever.” Ann. Intern. Med., 121:449-452, 1968.

KIDNEY ARTERIOVENOUS FISTULA SYNDROME

Syndromography

Clinic

Patients present the characteristic symptoms of high blood pressure, such as sometimes throbbing headache, tendency to dizziness, ringing in the ear, vision of daylight.

The early symptoms of heart failure are associated with these.

Physical examination. Arterial hypertension, audible noise especially in the upper anterior or posterior abdomen, palpable thrill, cardiomegaly, edema and other signs of heart failure.

Paraclinical examinations

Excretory urogram.

Renal arteriography. Allows checking the fistula.

Blood. Polycythemia.

Syndromogenesis and etiology

It is due to an arteriovenous fistula of the kidney.

Literature

Kirby, C.K., et al.: *Arteriovenous fistula of renal vessels; cae report." Surgery, 37: 267-271, 1955.
Scheifley, C.H.: “A new clinical syndrome producing hypertension arteriovenous fistula of the kidney." JAMA., 174:1625-1627, 1960.

FRALEY SYNDROME

Synonymy

S. compression of the calyx of vascular origin.

Syndromography

Clínic

It is characterized by intermittent unilateral flank pain, sometimes colicky. The pain intensifies on pressure and percussion of the renal region.

Sometimes it is accompanied by hematuria. Pyelonephritis and lithiasis in the upper calyx are common.

Paraclinical examinations

Urine. Microscopic or obvious hematuria.

Radiology. Decrease of the superior calyx with dilation of the middle calyx. Contrast arrest in the superior calyceal system. These alterations are more evident if the abdomen is compressed in the Trendelenburg position.

Angiography. An anomalous branch of the renal artery or vein is seen compressing the pelvic infundibulum. It is also observed in renal ptosis.

Syndromogenesis and etiology

The pain is probably due to periodic hyperdistension of the superior calyx. Alterations of the pelvic infundibulum are caused by extrinsic compression due to a branch of the renal artery or vein, which frequently coexists with renal ptosis.

Literature

Fraley, E.E.: "Vascular obstruction of superior infundibulum causing nephralgia: a new syndrome." New. Engl. J. Med., 275:1403-1409, 1966.

GLYCYNURIA SYNDROME

Synonymy

S. of idiopathic glycinuria.

Syndromography

Clinic

It is characterized by nephrolithiasis, but the stone is made of oxalate and contains 0.5% glycine. This occurs when the elimination of glycine through urine exceeds the saturation limit.

It is accompanied by the painful and septic symptoms typical of nephrolithiasis.

Paraclinical examinations

Blood. Normal plasma glycine level.

Urine. Hyperglycinuria.

Syndromogenesis and etiology

Probably dominant hereditary condition with a defect in the tubular transport system, especially glycine reabsorption.

Literature

De Vries, A.; S. Kochwa, et al.: "Glycinurias, a hereditary disorders associated with nephrolithiasis." Am. J. Med., 23:408-415, 1957.

HEMODIALYTIC SYNDROME

Syndromography

Clínic

The clinical picture is triggered abruptly after hemodialysis, especially renal, especially in cases of acute renal failure, acute nephritis and cortical necrosis of the kidney.

A light form is distinguished, without alterations in consciousness, which is manifested by muscle disorders and temporary disorientation, and a severe form, which begins with headache.

and vomiting followed by disorientation or loss of consciousness that can lead to death. Sometimes convulsive attacks and hypertension.

Paraclinical examinations

Blood. Hyperazotemia before hemodialysis that falls rapidly during it, but maintaining high nitrogen components in the cerebrospinal fluid.

Urine. The alterations typical of kidney injury.

Syndromogenesis and etiology

A high protein intake can cause an increase in urea in patients with significant kidney injuries, and although during dialysis urea decreases rapidly in the blood, in the cerebrospinal fluid the decrease is very slow due to alterations in the blood-brain barrier. This leads to cerebral edema and sometimes subdural hemorrhage.

Literature

Kennedy, A.C.; TO THE. Linton, et al.: "Urea levels in cerebrospinal fluid after haemodialysis." Lancet, 1:410-411, 1962.

HEMOLYTIC-UREMIC SYNDROME

Synonymy

S. de Gasser-Karrer. S. de Gasser.

Syndromography

Clinic

This syndrome occurs mainly in children, but cases in adults are becoming more frequent, with greater severity and mortality. In most cases, it begins with a viral illness, characterized by fever and general, respiratory and digestive disorders (diarrhea, sometimes bloody, vomiting and abdominal pain).

Bleeding may appear on the skin. This is followed by a picture of acute anemia with severe renal manifestations, alterations of the central nervous system and gastrointestinal hemorrhages. There is marked oliguria that can lead to anuria and hemoglobinuria in many cases. In adults, prodromal symptoms that resemble a viral disease are less noticeable, but have been related to pregnancy, postpartum, oral contraceptives, and infections that include: typhoid fever, gram-negative bacteremia, mumps, and infectious mononucleosis. Ischemic necrosis can affect many organs causing: heart failure and arrhythmias, jaundice, hepatosplenomegaly, pancreatitis and respiratory disorders.

serious.

Paraclinical examinations

Blood. Severe anemia, thrombocytopenia, decreased prothrombin time and accumulation of fibrin degradation products in serum. Uremia and creatinine retention, as well as other alterations typical of renal failure.

Kidney biopsy. Cortical necrosis and in immunofluorescence studies there is intense fibrin staining with the presence of antibodies in the membranes.

Syndromogenesis and etiology

The syndrome begins with hemolysis due to the fragmentation of erythrocytes, which is associated with thrombocytopenia and biochemical and pathological aspects of intravascular coagulation. This causes coagulation alterations and fibrin deposits in the small arteries and arterioles, with microangiopathic lesions and kidney necrosis and other organs.

The etiology is unknown.

Literature

Gianantonio C.A., et al.: "The hemolytic-uremic syndrome."
Nephron. 11:174, 1973.
Gasser, C.; E. Gauthier, et al.: "Hemobytisch-uremische Syndrome. Bilateral Nierenrindennekrose bei akuten erworbenen
hemolytischen Anmien". Schweiz. Med. Wschr., 85:905,
1955.

PRERENAL INSUFFICIENCY SYNDROME

Synonymy

S. of prerenal uremia. S. de Blum. S. de Blum-Van Caulaert. S. of hypochloremic azotemia. S. of extrarenal azotemia. S. of renal anoxia.

Syndromography

Clinic

It is characterized by asthenia, anorexia, nausea, dyspnea, progressive lethargy that leaves the patient exhausted and thirsty, then seizures appear and they fall into a coma.

Physical examination. Oliguria, irregular heart rhythm, tachypnea and signs of pulmonary edema are observed.

Paraclinical examinations

Blood. Anemia if the cause is blood loss.
Leukocytosis, thrombocytopenia, uremia, hyperphosphatemia, increased creatinine and uric acid, hyponatremia and decreased prothrombin consumption.

Urine. Scant, increased density, albuminuria.

Electrocardiogram. Signs of hyperkalemia.

Syndromogenesis and etiology

This syndrome is due to a decrease in renal perfusion caused by:

1. Decrease in cardiac output.
2. Blood loss.
3. Arterial hypotension.
4. Severe dehydration.
5. Post-surgery.
6. Post-trauma.
7. Inappropriate ADH secretion.
8. Vasoconstrictor drugs.
9. Instrumentations in the gastrointestinal tract.

Literature

Starling, E.H.: The Fluids of the Body. In The Herter Lectures, Chicago, Keener, 1909, p. 106.
Strauss, M.B. and L.G. Welt: Diseases of the Kidney, Boston, Little Brown, 1963.

KIMMELSTIEL-WILSON SYNDROME

Synonymy

S. of diabetes-nephrosis. Intercapillary glomerulosclerosis S. of diabetes-glomerulohyalinosis. S. of diabetic nephroangiopathy.

Syndromography

Clinic

It is generally observed in diabetes in young people treated with insulin and begins after 10 years of the disease. It is fundamentally characterized by high blood pressure, fluid retention with a clinical picture of nephrotic syndrome and later chronic renal failure with all its characteristics, death is fundamentally due to it.

In these patients, arterial and retinal lesions typical of the complicated diabetes phase are present.

Paraclinical examinations

Blood. In addition to glycemic alterations, elevations in urea and creatinine are seen in these cases.

When this happens, most patients die before 2 years after this alteration occurs. The electrolyte alterations characteristic of renal failure are also present.

Urine. Microalbuminuria is common in diabetics and ranges between 30 and 300 mg in 24 hours, but it can increase and become intense; If it exceeds 3 g/day, these patients do not live more than 6 years.

Kidney biopsy. The two most typical patterns can be seen: diffuse or nodular form that may or may not coexist. The diffuse form, more common, presents widening of the glomerular basement membrane, along with generalized mesangial thickening. In the nodular form, there is accumulation of PAS-positive material in the periphery of the glomeruli, a Kimmelstiel-Wilson lesion. In addition, there may be hyalinization of the afferent and efferent glomerular arterioles, "droplets" of Bowman's capsule, fibrin deposits, and occlusion of the glomeruli.

Syndromogenesis and etiology

The etiology is unknown and is considered related to insulin-dependent diabetes, according to animal experimentation and clinical observation. It has been observed that if a healthy kidney is transplanted into these diabetics, nephropathy occurs within a few years. If kidneys are transplanted from diabetics with renal syndrome to healthy people, nephropathy can improve. No immunological phenomena have been demonstrated in this syndrome.

Kidney transplant has significantly improved the future of these patients.

Literature

Mogensen C.E.; .K. Christensen: “Predictin diabetic nephropathy in insulin-dependent patients.” N. Engl. J. Med., 311:89, 1984.

Kimmestiel, P. and C. Wilson: "Intercapillary lesions in the glomeruli of the kidney." Am. J. Path., 12:83-98, 1936.

LIDDLE SYNDROME

Synonymy

S. of pseudoaldosteronism. S. of pseudohyperaldosteronism. S. of hypertension-hypokalemic alkalosis.

Syndromography

Clínica

This syndrome is characterized by polydipsia, polyuria, inability to concentrate urine. Severe arterial hypertension. Headaches and cerebral hypertensive crises, with paresthesias and papillary stasis. When it occurs in children, growth is normal.

Paraclinical examinations

Urine. Decreased density and low concentration power. Excretion of more than 80 mEg of potassium per day. Decreased sodium excretion.

Blood. In contrast to potassium loss, aldosterone levels are decreased and do not increase despite sodium deprivation and increased potassium stores.

Renin levels are normal or elevated as opposed to hyperaldosteronism, in which renin levels are low.

Triamterene, which blocks the exchange of potassium for sodium at the tubular level, is effective and corrects arterial hypertension and hypokalemic alkalosis, and also reduces potassium loss and increases sodium excretion.

Syndromogenesis and etiology

It is transmitted by autosomal dominant inheritance. The mechanism appears to be a defect in potassium transport across the renal tubule membrane.

The etiology is unknown.

Literature

Liddle, C.W. et al.: A familial renal disorder simulating primary aldosteronism but whit negligible aldosterone secretion. In Baulieu, E.E. and Robel, P (eds): Aldosterone, A Symposium. Oxford: Blackwell Scientific Publications, 1964, p. 352.

Royer, Nabib, Mathieu and Broyer: Pediatric Nephrology. Ed. Flammarion, Paris, 1973, p. 43.

BALKAN NEPHRITIS SYNDROME

Synonymy

Chronic nephropathy endemic to Yugoslavia. Nephropathy endemic to southeastern Europe.

Syndromography

Clinic

There may not be any renal symptoms such as edema, hematuria, or a history of upper respiratory infection.

Physical examination. The examination shows that the blood pressure is normal, only sometimes it is elevated.

Paraclinical examinations

Urine. Proteinuria no more than 1 g in 24 hours. Light leukocyturia and sometimes

Blood. Hyperazotemia (especially in patients over 30 years of age). Hyperchloremic acidosis.

Syndromogenesis and etiology

This syndrome affects a third of the population in endemic areas (small villages along rivers in Yugoslavia, Bulgaria, Romania). People who leave the place do not develop the syndrome, but those who live there develop the condition in approximately 10 years.

The etiology is unknown.

Literature

Hall, P.W.: G.J. Dammin; R.C. Griggs, et al.: "Investigation of chronic endemic nephropathy in Yugoslavia." Amer. J. Med., 39:210-217, 1965.

NEPHROTIC SYNDROME

Syndromography

Clinic

Nephrotic syndrome is a clinical term used to describe a condition characterized by severe proteinuria (more than 3.5 g per 1.75 m² of body surface area per day), low plasma albumin concentration, edema, and usually hypercholesterolemia. The cardinal characters can be associated with any disease in which the damage to the glomerular capillaries is of such intensity that there is a considerable and persistent increase in glomerular permeability and the consequent massive escape of albumin and other circulating low molecular weight proteins from the blood into the urine.

The most common symptom is edema, which can vary in intensity from slight morning periorbital dullness to anasarca and/or pleural effusions and ascites. Paleness of the skin in relation to edema and there are often white stripes in the nail bed due to chronic hypoalbuminemia.

Hypertension is found in up to 30% of cases, especially in adults with minimal glomerular lesions.

Other symptoms depend on the etiology of the syndrome (diabetes, lupus erythematosus, polyarteritis, etc.).

Paraclinical examinations

Urine. Proteinuria > 3 g, which can reach 15 g in 24 hours. In many patients there may be lipid substances in the sediment in the form of oval fatty bodies and bireflective bodies, which correspond to crystals of cholesterol esters.

Blood. Hypoalbuminemia, anemia and in many patients hyperipidemia. The most common pattern is: elevation of total cholesterol, triglycerides and lipoproteins a and

Kidney biopsy. It shows the membranous lesions in the glomerulus and its complete study (optical, immunofluorescence and electron microscopy) can specify the etiology of the syndrome.

Syndromogenesis and etiology

The conditions and/or causes that can cause or be associated with nephrotic syndrome are multiple.

The mechanism of edema production is related to the initial glomerular injury that causes proteinuria followed by hypoalbuminemia and a decrease in oncotic pressure, which causes a decrease in plasma volume. As a compensatory mechanism, there is an overproduction of aldosterone and antidiuretic hormone, which retain sodium and water, which produces more and more edema. There also appears to be less production of the so-called natriuretic hormone or auriculin. The cause of hyperlipidemia in this syndrome is uncertain, despite the many theories stated.

FAMILIAR JUVENILE NEPHROTISIS SYNDROME

Synonymy

S. de Fanconi I.

Syndromography

Clinic

This syndrome has been reported in children and young adults in both sexes. It is characterized by polydipsia, polyuria, blindness night followed by progressive decrease in the visual field and finally blurred vision.

Physical examination. In the eyes: effacement of the retinal arterioles, pallor of the papilla, deposits of yellow pigment in the retina, macular degeneration and opacity of the lens. Blood pressure is generally normal until the most advanced stages of the disease.

Paraclinical examinations

Urine. Decreased density, moderate proteinuria and minimal hematuria. Cultivation, negative. Abnormality in creatinine tolerance and phenolsulfonphthalein elimination.

Pyelography. Reduction in the size of the kidneys.

Blood. Uremia.

Kidney biopsy. Thinning of Bowman's capsule with hyalinization of the glomerulus, thinning of the membrane of the tubules. Tubular atrophy and interstitial fibrosis.

Syndromogenesis and etiology

It is a syndrome of genetic etiology of autosomal recessive inheritance.

McKusich identifies two independent entities with the same inheritance: juvenile familial nephronophthisis (MIM 256100) and renal dysplasia and retinal aplasia or S. Loken-Senior or S. renal-retiniano (MIM 266900). The basic defect is not clear.

Literature

Fanconi, G.: E. Hanhart; A. von Albertini, et al. : *Die familiare 'juvenile Nephronophthise'. Helv. Pediat. Acta, 6:1-49, 1951.

Meier, D.A.; J.W. Hess: “Familial nephropathy with retinitis pigmentosa.” Amer. J. Med., 39:58-69, 1965.

PROSTATIC OBSTRUCTION SYNDROME IN WOMEN

Synonymy

S. of Marion. (See S. of the bladder neck.)

Syndromography

Clinic

This syndrome is characterized by dysuria, which can range from mild discomfort to complete retention of urine.

Physical examination. On examination, the bladder is seen to be distended.

Paraclinical examinations

Urine. Discrete albuminuria or granulocytes in different stages of degeneration (pus).

Cystogram. It shows a filiform defect or narrowing in the neck, in the region of the internal orifice, similar to that observed in men due to prostate compression. The bladder wall presents different degrees of trabeculation.

Syndromogenesis and etiology

This syndrome is considered due to inflammation and/or hypertrophy of the group of glands that surround the posterior part of the urethra in women.

However, not all authors agree with this pathogenesis. But the rarity of obstructions in that part in

in women, due to granulomatous inflammation, neurological lesions, or cystocele, make it feasible. Folsomy O'Brien have demonstrated from an anatomopathological point of view the characteristics of the tissue extracted in these patients.

Literature

Caulk, J.: "Contracture of the bladder neck in the female." J. Urol., 6:341-343, 1921.

Marion, G.: De l'hypertrophie congenitale du col vesical". J.Urol. M d. Chil., 23:97-101, 1927.

Folsom, A, and H. O'Brien: "The female obstructing prostate." J.A.M.A., 121:573-580, 1943.

ORMOND SYNDROME

Synonymy

S. of periureteral fibrosis. S. of idiopathic retroperitoneal fibrosis. S. of fibrous retroperitonitis. S. of plastic periureteritis. of Gerota fasciitis.

Syndromography

Clinic

This syndrome is more common in men around 46 years of age, in women around 32 years of age. It is characterized by back pain of varying intensity, generally progressive, that radiates following the pattern of ureteral colic. Other times it is abdominal pain without a precise location. This pain can persist for months and sometimes radiates to the inguinal or genital region. It is frequently associated with:
nausea, vomiting, anorexia, malaise, fatigue, weight loss and constipation or diarrhea.

Sometimes it is associated with an ADH-resistant diabetes insipidus syndrome and in these cases the following occurs: dysuria, nocturia, oliguria, back pain and headache, edema and thirst.

Physical examination. It provides few elements: a slight increase in blood pressure and sometimes fever; Occasionally a tender mass can be palpated at the costovertebral angle.

Paraclinical examinations

Blood. Constant anemia, elevation of urea and erythrocyte sedimentation rate.

Pyelography. It allows us to specify the site of the obstruction in the middle ureter, almost always on the right, sometimes bilateral.

Syndromogenesis and etiology

It is possible that it is fasciculitis or a collagenous disease. Various causes have been thought of: medication abuse, relationship with other diseases (peritonitis, ulcerative colitis, lupus erythematosus, surgical interventions). See sclerosing lipogranulomatosis syndrome.

The etiology is unknown.

Literature

Ormond, J.K.: “Bilateral urethral obstruction due to envelopment and compression by an inflammatory retroperitoneal process.” J. Urol., 59:1072-1079, 1948.

Knowlan, D.; M. Corrado; G.E. Schreiner, et al.: Periureteral fibrosis with diabetes insipidus-like syndrome occurring with progressive partial onstruction of ureter unilaterally". Amer. J. Med., 28:22-31, 1960.

OSTERTAG SYNDROME

Synonymy

S. of hereditary amyloidosic nephropathy.

Syndromography

Clínic

It affects both sexes, variable age of onset, arterial hypertension, large hepatosplenomegaly, hematuria, edema (clinical expression of nephritis or nephrosis).

Paraclinical examinations

Blood and urine. Evidence of nephropathy. Congo red test.

Kidney biopsy.

Syndromogenesis and etiology

Of genetic etiology and autosomal dominant inheritance.
It is also known as type III amyloidosis.

Literature

Ostertag, B.: "Familiare Amyloid-Erkrankung." Z. Menschl. Vereb Konstit Lehre, 30;105-115, 1950.

PEYRONIE'S SYNDROME

Synonymy

S. of plastic induration of the penis. S. of fibrous cavernositis.S. by Van Burens.S. of chronic indurative cavernositis.

Syndromography

Clínic

This syndrome is observed in middle age and in some cases in older men. It is characterized by a slowly developing induration on the dorsum of the penis. At first this induration is elastic and later cartilaginous, which extends about 2-8 cm and then grows to the distal end of the penis. It is accompanied by pain and curvature of the penis; inability for erection in the region distal to the process and interference with intercourse, as the penis curves laterally or dorsally. On palpation, an irregular hardening is seen on the dorsum of the penis.

The condition is benign, but can lead to suicide due to the inability to have intercourse.

Paraclinical examinations

Blood. Syphilis should be ruled out by serology.

Biopsy. In this syndrome, lesions that resemble a keloid are observed, with the absence of inflammatory signs or elements that suggest a neoplastic lesion.

Syndromogenesis and etiology

The association of this syndrome with Dupuytren's syndrome is frequent, 30% of cases. It is considered a constitutional predisposition.

The etiology is unknown.

Literature

Peyronie, F. de la: "Sur quelques obstacles qui sópposent léjaculation naturelle de la semence." Academic Mem. Chir. Paris, 1:425, 1743.
Parton, I.: Urology in General Practice. London, Butterworth, 1960.

KIDNEY CYST SYNDROME

Synonymy

S. of cystic kidney. S. of polycystic disease. Adult polycystic kidney disease (PKD 1).

Syndromography

Two types are recognized: children and adults.

The infantile type is characterized by:

Clínica

Nausea, vomiting and dehydration.

Physical examination. Detected: abdominal distension, palpation of bilateral renal masses (except in the small cyst variety).

Physical signs of cysts may be found in other organs, such as the brain, lung, liver, pancreas; but they are usually subordinated to kidney lesions, which are what cause the main symptoms.

Paraclinical examinations

Urine. Albuminuria, hematuria and increased epithelial cells.

Blood. Hyperchloremic acidosis.

Imaging.

Pyelogram.

Ultrasound.

Renal scanning with radioactive isotopes.

The adult type is characterized by:

Clinic

It can be asymptomatic for several decades. Other times it is manifested by lower back pain, dysuria, hematuria with or without pain, weakness and an increase in the abdomen.

Physical examination. Generally polylobulated renoabdominal masses are palpated that correspond to the kidneys and that are prominent in the abdominal cavity. Arterial hypertension.

Paraclinical examinations

They are those indicated in the infantile type.

Syndromogenesis and etiology

It has genetic etiology. This mutation is transmitted as an autosomal dominant inheritance. The PKDl gene has been located on chromosome 16 p13.11, to p13.3.

The basic defect, although not well delineated, is suggested to be related to proton channel defects.

Literature

Gillespie, G. A. et al.: CpG island the region of an autosomal dominant polycystic kidney disease locus defines the 5-prime end of a gene encordiny a putative proton channel. Proc. Nat. Academic Sci., 88:4289-4293, 1991.

PRIMARY URETERAL REFLUX SYNDROME

Synonymy

S. of megaureter-megabladder. S. of aganglionic bladder. S. of idiopathic dilation of the urinary tract.

Syndromography

Clinic

It is characterized by repeated urinary infections in children, with vomiting, febrile episodes, and pyuria, which are temporarily cured with antibiotics. It can occur at older ages.

Physical examination. A very dilated, flaccid or tense bladder that can reach the navel can be percussed and palpated.

Paraclinical examinations

Cystography. It shows a very large bladder with very thin walls, but hypertrophic mucosa; marked reflux towards the ureters that are very dilated and also towards the renal calyces. The ureteral orifice in the bladder is wide and dilated. The urethra is normal.

Syndromogenesis and etiology

This is a primary ureteral reflux, which may be due to poor development of the trigone muscles or other causes. The so-called megabladder with very thin walls is secondary and is due to the compliance of the bladder wall in the child.

It is considered that it may be due to a functional adaptation to maintain sufficient evacuation, despite vesicoureteral reflux.

It is associated with congenital megacolon, which is why it has been suggested that it is due to an aganglionic disease or defects in the pelvic parasympathetic.

Literature

Kretschmer, H.L. and J. R. Greer: "Insufficiency at the ureterovesical junction." Surg. Gynec. Obstet., 21:228-231, 1915.
Paquin, A.J. Jr.; GO. Marshall; J.H. McGovern: "The megacystis syndrome." J. Urol., 83:634-646, 1960.

RENOY (JUHELL) SYNDROME

Synonymy

S. of bilateral necrosis of the renal cortex.

Syndromography

Clínic

In two-thirds of cases, this syndrome begins during pregnancy or the postpartum period in the form of eclampsia or preeclampsia. The clinical picture begins with the death of the fetus and its expulsion, in other cases it is associated with surgical trauma, infections or poisoning. It is manifested by bilateral lumbar pain and epigastric pain, vomiting. Sometimes loss of consciousness. Oliguria, hematuria and progression towards anuria, uremia and coma.

The prognosis is fatal, death occurs in a short time, with few cases surviving.

Paraclinical examinations

Urine. Hematuria, cylindruria and proteinuria.

Blood. Hyperazotemia, hyperkalemia, hypermagnesemia and hypochloremia.

Kidney biopsy. Multiple cortical necrosis with hyalinosis of the glomeruli.

Syndromogenesis and etiology

Experiments in animals have shown that bilateral renal cortical necrosis can be caused by different noxas, which determine a spasm at the level of the renal vessels (cholera or other bacterial toxins, diethylene glycol, dioxan, lithiumcarmine, neurohypophyseal hormones, adrenaline, serotonin, shock). However, although in humans some of these factors are repeated with analogous mechanisms, in pregnancy there are complex functional conditions and predisposing factors that facilitate a shock and that can act as triggering factors.

Literature

Renoy, J.: "De 1' anurie précoce scarlatineuse." Arch. gen. Med., 17:385-410, 1886.

Matlin, R.A. and N.E. Gary: "Acute cortical necrosis. Case report and review of the literature." Am.J.Med., 56:1 10-1 18, 1974.

THORN SYNDROME

Synonymy

S. of salt wasting nephritis. Pseudo Addisson

Syndromography

Clínica

This syndrome is characterized by marked asthenia, dizziness, fainting and muscle cramps. Digestive symptoms: anorexia, nausea and vomiting. In its evolution, a picture of prostration, arterial hypotension (at the beginning), followed by hypertension (due to increased aldosterone and hypokalemia), mental confusion, collapse and coma occurs.

Signs of nephropathy: polyuria, hyposthenuria, polydipsia.

Paraclinical examinations

Blood. Marked decrease in sodium and hypochloremia.

Increase in renin and later in aldosterone, followed by potassium deficiency. Hyperazotemia.

Urine. Marked increase in sodium excretion.

Syndromogenesis and etiology

It is generally observed in some pyelonephritis in which there is excessive loss of salt through the urine, due to a tubular inability to reabsorb sodium.

Simulates the clinical picture of Addison's disease.

Literature

Thorn, G. W. et al.: "Renal failure simulating adrenocortical insufficiency. " New England J. Med., 231:76, 1944.

HALF AND HALF NAIL SYNDROME

Synonymy

S. of azotemic onychopathy

Syndromography

Clinic

In chronic kidney disease with hyperazotemia, 25 cases out of 1, 500 present nail alterations. It can be seen that the nail bed of the nails has reddish or reddish-brownish bands that occupy half of the bed, but the other portion remains normal in color. The proximal half is white and the distal half has the indicated coloration. If venous return is prevented, the contrast between the two areas is not substantially altered. This pattern has no tendency to extend outside the nail.

There is also no relationship between the severity of uremia and nail changes.

Paraclinical examinations

Blood. Uremia and decreased creatinine clearance.

Syndromogenesis and etiology

It is observed in 20-40% of cases of chronic uremia.

They have sometimes been found in cases of cylindruria without uremia. It can also be associated with liver cirrhosis.

The etiology is unknown.

Literature

Bean, W.B., “A discourse on mail goth and unusual fingernails.” Trans. Amer. Clin. Climate Ass., 74:152-167, 1963.

Lindsay, P.G: "The half and half nail." Arch. Int. Med., 119:583-587, 1967.

CARDIA CA UREMIA SYNDROME

Syndromography

Clinic

This syndrome occurs in patients with chronic uremia, treated with a selectively low-protein diet. It can be seen: pronounced cardiomegaly, galloping rhythm and severe hypotension. Pericarditis and arrhythmias are common and show marked sensitivity to cardiac glycosides.

Paraclinical examinations

Blood and urine. They show the alterations typical of chronic uremia: low hematocrit that persists even after the clinical picture is controlled.

Chest x-ray. Cardiomegaly.

Electrocardiogram. It shows the type of arrhythmia that accompanies this

Syndromogenesis and etiology

It is considered a progression of the uremic condition or that it is a secondary process caused by diet, prolonged hypertension or chronic anemia.

The etiology is unknown.

Literature

Bailey, G.L., C.L. Hampers, J.P. Merill: Ann. Meeting of Amer.Soc. Artif. Internal Organs. Quoted by J.A.M.A., 200:8-30, 1967.

POSTGONOCOCCAL URETHRITIS SYNDROME

Syndromography

Clinic

Symptoms appear within 20 days after treatment with penicillin and probenecid in patients with gonococcal urethritis, who have not had new contamination.

It is characterized by urethritis with the expulsion of pus from the urethra without the previous germs.

Paraclinical examinations

Smear and cultivation. They show mycoplasma infection. This does not happen when patients are treated with tetracycline.

Syndromogenesis and etiology

It is due to a mycoplasma infection associated with gonorrhea that is exacerbated after the administration of penicillin.

Literature

Holmes, K.K., D.W. Johnson, T.M. Floyd, et al.: "Studies of venereal disease. II. Observations on the incidence, etiology, and treatment of the postgonococcal urethritis syndrome."
J.A.M.A., 202:467-473, 1967.

RELATED KIDNEY SYNDROMES

Syndromes with abnormal urine color

Dark brown or black.

- Blackwater fever.
- Bywaters.
- Disseminated melanotic sarcoma.
- Garrod.
- Hemoglobinuria.
- Tyrosinosis and tyrosinuria.

Pale color.

- Diabetes mellitus.
- Diabetes insipidus.

Red.

- Hematurics.
- Hemoglobinurics.
- Monday morning disorder.
- Myoglobinuria.
- Porphyrias.
- Red diaper.

Green and blue color.

- Drugs.
- Chronic obstructive icterus.
- Blue diaper.

Nephrocutaneous syndromes

They comprise a group of diseases in which skin and kidney lesions are present, or skin manifestations and excretion of abnormal substances in the urine.

Hereditary.

- Bourneville.
- Ehlers-Danlos.
- Fabry.
- Gelfarb-Hyman.
- Grönblad-Strandberg-Touraine.
- Herrick.
- Malformation of the familial urogenital tract-ear.
- Marfan.
- Potter.
- Pringle-Bourneville.
- Rendu-Osler-Weber.
- Urticaria pigmentosa.
- Von Hippel-Lindau.
- Von Recklinghausen.

Vasculitis and collagen disorders.

- Dermatomyositis.
- Scleroderma.
- Goodpasture.
- Kussmaul-Maier.
- Systemic lupus erythematosus.
- Schönlein-Henoch.
- Stevens-Johnson.
- Allergic vasculitis.
- Wegener.

Metabolic.

- Amyloidosis.
- Calcinosis.
- Hereditary amino acid deficiency.
- Folling.
- Drop.
- Alkaptonuric ochronosis.
- Porphyria.
- Erythropoietic protorphyria.

Neoplastics.

- Kahler-Bozzolo.
- Kaposi.

Miscellaneous.

- Cold agglutination.
- Besnier-Boeck-Schaumann.
- Dysproteinemias.
- granulomatous diseases.
- Infections.
- By radiation.
- Post-transfusion purpura.
- Waldenstrom.

Due to corticosteroids.

- Cushing.
- For antibiotics.
- Enterocolitis due to antibiotics.

THORACIC OUTLET SYNDROME

Synonymy

S. of neurovascular compression. S. of the costoclavicular gorge. S. costoclavicular. S. compression of the pectoralis minor muscle. S. subcoracoid-pectoralis minor.

Syndromography

Clínic

It is manifested by pain in the chest, shoulder, arm, hand. Numbness. Color changes in the skin. Claudication. Neurovascular symptoms in the upper limb after prolonged hyperabduction of the arm during physical activity or sleep. Symptoms appear when the upper limbs are circumducted and the hands are clasped above the head.

Physical examination. Noise in the supraclavicular or infraclavicular fossa. Weak pulse, decreased blood pressure on the affected side. Atrophy of hand muscles, weakness and atrophy in the forearm.

Paraclinical examinations

Arteriography. Shows the site of arterial obstruction (usually second portion of the subclavian). Sometimes radioisotopes demonstrate flux reduction.

Radiology. Osteoporosis of the phalanges (in patients with embolism). Bone anomalies: supernumerary rib, anomaly of the first cervical vertebra and its corresponding rib, clavicular deformity, previous thoracoplasty.

Syndromogenesis and etiology

Hyperabduction of the upper limb compresses the neurovascular elements at the crossing point of the coracoid process and the head of the humerus.

Literature

Cantrell, J.R., et al.: *A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart." Surg. Gynecol. and Obstet.. 107:602, 1958.

Wright, I.S.: "The neurovascular syndrome produced by hyperabduction of the arms." Amer. Heart J., 29:1-19, 1945.

Hollander, J.L.: Arthritis and Allied Conditions. ed. 7, Philadelphia, Lea & Febiger, 1966.

AYERZA SYNDROME

Synonymy

S. de Ayerza-Arillaga. S. cardiac blacks.

Syndromography

Clínic

Symptoms appear gradually in patients around the fifth decade of life and who have a history of bronchopulmonary symptoms.

It is characterized by symptoms of respiratory failure with cough and severe dyspnea. Sleepiness.

Physical examination. Marked cyanosis, especially on the face, hands and feet. Signs of pulmonary emphysema and right ventricular hypertrophy of the heart. Hepatomegaly, hepatojugular venous reflux, lower limb edema, especially when right heart failure syndrome appears.

Paraclinical examinations

Blood. Polycythemia.

Respiratory functional tests. Pathological.

Chest radiology. With signs of emphysema and right ventricular hypertrophy of the heart. Arterial atheromatosis with narrowing of the vascular lumen.

Syndromogenesis and etiology

The pathogenesis of the syndrome is associated with diseases of the pulmonary arteries such as atherosclerosis, syphilis, etc.

The etiology is unknown.

bibliography

Ayerza, L.: “Maladie d'Ayerza, sclérse secondaire de 1'artere pulmonaire (cardiaques noirs)". Sem. Med. B. Aires, 32:43, 1925.

J Ayerza, L.: Unpublished clinical lecture at National University of Buenos Aires, 1901.

Wintrobe, M.M.: Clinical Hematology. Philadelphia, Lea & Febiger, 1967.

BAGAZOSIS SYNDROME

Syndromography

Clinic

It occurs in workers in the sugar industry who have to do with sugarcane bagasse. It can be acute, subacute or chronic. Of insidious or sudden appearance due to exposure to bagasse dust. Fever, chills, sweating, dry cough, anorexia, weight loss, chest pain and occasionally hemoptysis.

Physical examination. Tachypnea, tachycardia, signs of hypoxia. Thin rales on auscultation.

Paraclinical examinations

Chest radiology. Increase in the reticular fabric of the lung. Fine nodular images.

Blood. Moderate anemia. No eosinophilia. Sometimes monocytosis. Positive skin reaction against specific antigen. Interstitial pneumonitis.

Syndromogenesis and etiology

Hypersensitivity to sugarcane bagasse dust.

Literature

Jamison, S.C., and J. Hopkins: "Bagassosis, a fungus disease of the lung case report." New Orleans Med. Surg. J., 93:580-582, 1941.

Nicholson, D.P.: "Bagasse worker's lung." Amer. Rev. Resp. Dis., 97:546-560, 1968.

CARSSON SYNDROME

Synonymy

Carsson's disease. Decompression S. Air decompression sickness. Aeroembolism.

Syndromography

Clinic

It occurs in workers who carry out their work in submersion chambers, tunnels, under high pressure. Upon returning to atmospheric pressure, they suffer from headache, asthenia, nausea, vomiting, dizziness, dyspnea, shock, and dry cough. Paraplegia with intestinal paralysis and urinary retention, seizures, abdominal and joint pain may occasionally occur.

Physical examination. Tachypnea with shallow respirations. Shock, hypotension and bradycardia.

Syndromogenesis and etiology

The cause of this syndrome is the increase and release of circulating nitrogen in the blood and its migration to the different tissues due to sudden decompression.

Literature

Bassoe, P.: "The late manifestations of compressed air disease", Amer. J Med Sci. 165:526-542. 1913.

Grinker, R.R. and A. L. Sahs: Neurology. ed. 6, Springfield, Thomas, 1966.

CEELEN-GELLERSTEDT SYNDROME

Synonymy

S. of idiopathic pulmonary hemosiderosis. S. of idiopathic dark induration of the lungs.

Syndromography

Clinic

It is more common in men in early stages of life. Sudden presentation, recurrent episodes of cough attacks, dyspnea, hemoptysis, sometimes fever and abdominal pain.

Physical examination. Skin pallor, sometimes jaundice and cyanosis, clubbing of fingers and splenomegaly.

Paraclinical examinations

Blood. Hypochromic anemia, hyperbilirubinemia, normal coagulation test. Late polycythemia.

Chest radiology. Poorly defined lung opacity. Pattern of magnification of the pulmonary reticulonodular pattern. Cardiomegaly due to chronic corpulmonale in some cases.

Syndromogenesis and etiology

It is considered a variant of Goodpasture syndrome, especially in adults.

The etiology is unknown.

Literature

Bronson, S.M.: "Idiopathic pulmonary hemosiderosis in adults: Report of a case and review of the literature". Am.J. Roentgenol., 83:260, 1960.

Ceelen, W.: Die Kreislaufstoe-rungen der lungen. In: Henke-lubarsch: Handbuch der Speziellen pathologischen Anatomie und Histologie, v. 3, Berlin, Springer, 1931, p.10.

Gellerstedt, N.: "Uber die 'essentielle' anamisierende form der braunen lungeninduration." Acta Pathol. Microbiol. Scand., 16:386, 1939.

Soergel, K.H. and S.C. Sommers: "Idiopathic pulmonary hemosiderosis and related syndrome", Amer. J. Med.. 32:499- -511, 1962.

CERVICAL SYNDROME

Synonymy

S. compression of cervical nerve roots. Cervical spondylitis. S. of Rust. S. of the cervical rib. S. of hyperadduction. S. of the first supernumerary rib.

Syndromography

Clinic

This syndrome almost always appears in adulthood, after trauma to the shoulder due to stretching or straining the arm or pregnancy. It is generally unilateral, although supernumerary ribs can be bilateral and more frequently in women. There is pain in the ulnar border that increases with movements, weakness of the hands, arm, shoulders, numbness and loss of sensation.

The patient presents pain or sensory, motor and trophic changes along the path of the nerves taken, accompanied by limitation of neck movements, loss of balance, ringing in the ears, nausea, vomiting, tachycardia and circulatory changes.

Physical examination. There is evidence of: cyanosis, edema, coldness, vasomotor spasms, muscle atrophy, weak pulse on the affected side, occasionally a bone mass is palpable in the supraclavicular fossa on that side.

Added discomfort and muscle spasm in the affected areas and muscular dystrophy. Reflexes may be normal, decreased, or absent. Pupil dilation and other signs of Horner syndrome sometimes. Difference in blood pressure between both members.

Paraclinical examinations

Radiology. Shows the cervical supernumerary rib, it must be differentiated from the cervical disc.

In the spinal column, osteophyte formation, degenerative changes and narrowing of the intervertebral spaces are observed.

Syndromogenesis and etiology

The etiology can be tumorous or inflammatory.

It is produced by the supernumerary cervical rib, which irritates the brachial plexus, compressing it between the rib and the anterior scalenus muscle. Due to the familial nature, a genetic etiology with autosomal dominant inheritance has been proposed.

A similar syndrome also occurs due to compression and/or irritation of the roots within the foramen or canal before the division of the nerve trunk. In these cases it may be due to a tumor or inflammatory cause.

Literature

Holst, S.: "Cervical rib and associated vascular complications." J. Oslo City Hosp., 13:173-182, 1963.

Jackson, R.: The cervical syndrome. Springfield, Thomas, 1958.

Jackson, R: The Syndrome of Cervica Nerve Root Compression. in Hollander, J.L.: Arthritis and Allied Conditions. Philadelphia, Lea & Febiger, 1966.

Lishman, W.A. and W.R. Russell: The barchial neuropathies. Lancet, 2:941-946, 1961

Murphy, T.: "Brachial neuitis caused by pressure of first rib." Aust. Med. J., 15:582-585, 1910.

Schapera, J.: "Autosomal dominant uninheritance of cervical ribs." Clin. Genet. 31:386-388, 1987.

Smyth, C.J., ed.: "Rheumatism and Arthritis." Ann. nt. Med., 59 suppl. 4, 74-75, 1963.

Willshire: "Supernumerary first rib. Lancet, 2:633, 1860.

CYANOTIC SYNDROME WITH FINGERS IN DRUMSTICK AND HEPATOPATHIES

Synonymy

Cyanotic S. with clubbing of fingers and polycythemia.

Syndromography

Clinic

It occurs in both sexes and all ages, most frequently in children.

Physical examination. Cyanosis with or without dyspnea. Clubbing fingers. It coexists with liver cirrhosis, with the characteristics of this

Paraclinical examinations

Blood. Secondary polycythemia.

Laparoscopy. Nodular liver signs of portal hypertension. Low arterial oxygen saturation evidence of arteriovenous shunts.

Chest radiology. It does not reveal pulmonary alteration.

Syndromogenesis and etiology

The release of a substance in the liver that causes pulmonary arteriovenous shunts in individuals with a familial predisposition is proposed.

The etiology is not completely clarified.

Literature

Fluckiger, M.: Vorkommen von trommelschlagel-formigen Fingerendphalangen ohne chronische Veranderungen an den Lungen oder am Herzen". Wien. Me. , 34:1457, 1884.

Silvermann, A.; M.D. Cooper; J.H Moller, et al.: "Syndrome of cyanosis, digital clubbing, and hepatic disease in siblings." J. Pediat.. 72:70-80, 1968.

CITELLI SYNDROME

Synonymy

S. of aprosexia.

Syndromography

Clinic

It occurs in children and is characterized by loss of concentration power, insomnia, drowsiness, and mental retardation.

Physical examination. Facial changes may occur due to upper respiratory obstruction.

Paraclinical examinations

Ororhinopharyngeal examination. Large adenoid masses are confirmed.

Radiology of perinasal sinuses. Opacity that in many cases reveals sinus inflammatory processes.

Syndromogenesis and etiology

It is due to rhinopharyngeal obstruction due to adenoid masses.

Literature

Citelli, S.: "Vegetazioni adenoidi e deafmutism." Boll. d. evil. d. orecchio, d. gola e.d. naso, 22: 141-150, 1904.

DYSGAMMAGLOBULINEMIA TYPE I SYNDROME

Syndromography

Clinic

It is more common in males and occurs at any age. The frequency is greater in childhood after the sixth month of life. It is characterized by repeated bacterial respiratory diseases, pneumonia, sinusitis, rheumatoid arthritis, and an increased incidence of lymphoma and leukemia.

Physical examination. Hepatosplenomegaly and lymphadenopathy.

Paraclinical examinations

Blood. Hypogammaglobulinemia (IgA and IgG deficiency. lgM is normal or increased). Decreased cellular immunity.

Radiology. Pneumonic condensations, atelectasis, bronchiectasis, absence of hilar lymphadenopathy.

Pathology. Normal number of plasma cells present in various tissues.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Barth, W.F.: "An antibody deficiency syndrome." Amer. J. Med., 39:319-334, 1965.

Christian, J.C., et al.: "Sisters with low birth weight, dwarfism. congenital anomalies, and dysgammaglobulinemia". Am.J. Dis. Child., 122:529, 1971.

Sell, S.: "Immunological diseases". Arch Path.. 86:95-107. 1968.

SCALENE SYNDROME

Synonymy

S. of the cervical rib without cervical rib. S. of neurocirculatory compression by the

Syndromography

Clinic

It is characterized by paresthesias (corking), burning pains (sometimes) that extend especially through the territory of the ulnar nerve (ulnar nerve) caused by the compression of the cervical roots (CVII-CVIII). It is accompanied by tactile hyperesthesia. Motor disorders (pressure weakness) in the territory of the median nerve, with atrophy of the thenar or ulnar eminence (claw hand). Coldness of the hand and forearm, radial microphygmia and cyanosis (due to compression of the subclavian artery).

Syndromogenesis and etiology

This syndrome is due to isolated hypertrophy or fibrosis of the anterior scalenus. Abnormal development of the first rib.

PULMONARY ARTERIOVENOUS FISTULA SYNDROME

Synonymy

S. of congenital arteriovenous aneurysm or pulmonary varicose veins. of cavernous hemangioma of the lung.

Syndromography

Clinic

It occurs in young adult men, it can also be seen in infancy and young children. The symptoms depend on the size of the shunt; it may be asymptomatic or there may be dyspnea, chest pain, dizziness, loss of consciousness, Adam-Stokes syndrome and hemoptysis.

Physical examination. Cyanosis, clubbing of fingers and watch glass nails. On auscultation, a continuous murmur can be heard over the affected lung. In the skin and mucous membranes there may be associated hemangiomas.

Paraclinical examinations

Blood. Polycythemia.

Chest radiology. Increased lung density.

Fluoroscopy. Pulsatile masses or not.

Tomography. Rounded lesion linked to the vessels.

Angiocardiography. It can demonstrate arterial fistulas venous.

Syndromogenesis and etiology

In cases where Rendu-Osler syndrome is present, an inherited condition may exist.

The etiology is unknown.

Literature

Churton, T.: "Leeds and West-Riding Medico-Chirurgical Society: Multiple aneurism of pulmonary artery." Brit. Med. J., 1:223, 1897.

Friedberg, C.K.: Diseases of the Heart. ed. 3, Philadelphia, Saunders, 1966.

GOODPASTURE SYNDROME

Synonymy

S. of glomerulonephritis with pulmonary hemorrhage. S. purple lung. S. nephritic. S. of interstitial hemorrhagic pneumonitis with nephritis.

Syndromography

Clinic

It predominates in men, in an age range between 16-61 years, especially in the 20s. Every day more cases are reported in women. A fifth of cases present viral respiratory infections, hemoptysis, sometimes of great intensity, dyspnea, cough, easy fatigue, occasionally nausea and vomiting, weight loss, chest pain, hematuria, proteinuria, with progressive renal failure.

Physical examination. Pallor, rarely hypertension, edema, skin rash, splenomegaly. Hypertension and edema occur more frequently in the more advanced stages of the disease.

Paraclinical examinations

Blood. Normocytic and normochromic anemia, sometimes leukocytosis. Hyperazoemia at the end of the disease. In more than 90% of cases, circulating antibodies against glycopeptide antigens related to non-collagenous tissue are found in type IV collagen (basement membrane). Circulating antibody levels have diagnostic but not prognostic value. No immune complexes or cryoimmunoglobulins were detected.

lantes.

Urine. Proteinuria, hematuria, leukocyturia.

Chest radiology. Inflammatory infiltrate in the lung bases.

Sputum. Macrophages filled with hemosiderin. It can be confused with pulmonary hemosiderosis. By immunochemistry and fluorescence, deposits in the glomerular basement membrane of IgG compounds and other globulins are recognized in the kidney.

Kidney biopsy. The glomeruli can range from an almost normal appearance to focal proliferative glomerulonephritis with necrosis. There is frequently extensive extracapillary proliferation (crescent images). Immunofluorescence studies may reveal typical linear deposits and anti-basement membrane antibodies.

Syndromogenesis and etiology

The possible viral etiology and autoimmune factors are pointed out.

In the lung there are areas of intraalveolar hemorrhage. Increase in the thickness of the alveolar walls with collagenization. In the kidney there is progressive glomerular fibrosis. Interstitial inflammation with fibrosis, dilation and atrophy of the tubules.

The most current theory proposes an immunological process due to an antigen-antibody reaction, which damages the alveoli and the glomerular basement membrane.

The etiology is unknown

Literature

Brannan, H.M., et al.: "The roentgenographic appearance of pulmonary hemorrhage associated with glomerulonephritis." Am. J. Roentgenol., 90:83, 1963.

Eisinger, A.J.: "Goodpasture's syndrome: Failure of nephrectomy to cure pulmonary hemorrhage." Am. J. Med., 55:565, 1973.
Goodpasture, E.W.: "The significance of certain pulmonary lesions in relation to the etiology of influenza." Amer. J. Med. Sci., 158:863, 870, 1919.
Maddock R.K.; YOU. Stevens; K. Reemtsma, et al.: "Goodpasture syndrome; cessation of pulmonary hemorrhage after bilateral nephrectomy." Ann. Intern. Med., 67:1258-1264, 1967.
Proskey, A.J., et al.: "Goodpasture's syndrome. A report of a five cases and review of the literature." Am. J. Med., 48:162, 1970.

HAMMAN-RICH SYNDROME

Synonymy

S. of alveolocapillary block. S. of diffuse interstitial pulmonary fibrosis. S. of the rheumatoid lung. Cryptogenic fibrosing alveolitis S. S. of acute interstitial pneumonia fulminante.

Syndromography

Clinic

Insidious presentation, progressive dyspnea, rapid and shallow breathing, dry cough, accompanied by febrile attacks. During its evolution, dyspnea is permanent and cyanosis sets in.
Weight loss and easy fatigue.

Physical examination. Cyanosis, clubbing of fingers and watch glass nails, fine rales at the lung bases, right heart failure. Increased venous pressure.

Paraclinical examinations

Blood. High titers of rheumatoid factor in some cases. Decreased carbon dioxide tension in arterial blood.
Polycythemia.

Radiology. Fine diffuse and reticulated mottling at the beginning of the disease, honeycomb appearance at the end.

Respiratory function tests. There is no evidence of obstructive expiration syndrome. Reduction of the diffusion capacity of carbon dioxide. Thickening and diffuse fibrosis of the alveolar wall in the lung biopsy, with infiltration of mononuclear cells in alveolar spaces.

Syndromogenesis and etiology

In some cases it is possible it is due to the association with rheumatoid arthritis or scleroderma, exposure to radiation, contact with beryllium or asbestos.

In other cases it has been suggested that it is transmitted by autosomal dominant inheritance.

It is observed in all pathologies that reduce the alveolocapillary diffusion capacity such as: interstitial lung diseases, granulomatosis, scleroderma, alveolar cell carcinoma, sarcoidosis, alveolar proteinosis, miliary tuberculosis, pulmonary hemosiderosis, histiocytosis X.

The etiology is unknown.

Literature

Fishman, A.P.; GM. Turino and E.H. Bergofsky: "The syndrome of alveolar hypoventilation." Amer. J. Med., 23:333-339, 1957.
Fraser. R.G. and J.A.P. Paré: Diffuse Idiopathic Interstitial Pulmonary Fibrosis (Hamman-Rich syndrome), in Diagnosis of Diseases of the Chest (Philadelphia: W.B. Saunders Co., 1970), pp. 1 104-1109.
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Lemire, P., et al.: "Patterns of desquamative interstitial pneumonia (D.ILP) and diffuse interstitial pulmonary fibrosis (D.I.E)." Am. J. Roentgenol., 115, 479, 1972.
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Swaye, P.; H.S. Van Orstrand; L.J. Mc Cormack and S.E. Wolpaw: *Familial Hamman-Rich Syndrome, report of eight cases." Dis. Chest.. 55:7-12, 1969.

HEDBLOM SYNDROME

Synonymy

S. of primary acute diaphragmitis.

Syndromography

Clinic

The onset of this syndrome is sudden, with pain in the shoulders and back, as well as in the upper abdomen.

Physical examination. There is a decrease in respiratory expansion upon inspiration and a marked limitation on deep inspiration due to pain.

Paraclinical examinations

Chest x-ray and fluoroscopy. Decreased diaphragmatic mobility. In some cases there are inflammatory signs in the pleura and peritoneum.

Syndromogenesis and etiology

A nonspecific myositis of one or both hemidiaphragms is observed with leukocyte infiltration of the diaphragm and thickening.
ment of muscle fibers.

Literature

Joannides, M.: "Acute primary diaphragmitis (Hedblom's syndrome)". Dis. Chest., 12:89-110, 1946.

HPOVENTILATION SYNDROME

Synonymy

S. of hypercapnia with papilledema. S. of carbon dioxide narcosis.

Syndromography

Clínic

This syndrome has two forms:

Mild form: headache, nausea and anorexia.

Severe form: delirium. If hypoxia exists, there is paleness, cyanosis or shock. Delirium may persist in some cases depending on hypoxia.

Physical examination. Respiratory depression in frequency and depth. Patients with respiratory failure are tachypneic with hypoventilation. Papilledema.

Paraclinical examinations

Blood. Determination: pH and serum bicarbonate; arterial oxygen tension, hemoglobin, blood glucose. Increased tension of the cerebrospinal fluid.

Syndromogenesis and etiology

This syndrome can be caused by different causes:

respiratory acidosis, metabolic alkalosis, hypokalemic alkalosis, alkalosis due to bicarbonate ingestion, intoxication by depressant drugs, head trauma, Pickwick syndrome, cardiac diseases, diuretic therapy, Cushing's syndrome, Conn's syndrome, myxedema coma.

Literature

Miller, A.; R.A. Bader; M. Bader: "The neurological syndrome due to marked hypercapnia with papilledema." Amer. J. Med., 33:309-318, 1962.

Stemmer, E.A.; D.W. Crawford; J.W. List, et al.: "Diaphragmatic pacing in the treatment of hypoventilation syndrome." J. Thorac. Cardiov. Surg., 54:649-657, 1967.

CHRONIC HYPOVENTILATION SYNDROME

Synonymy

S. of emphysema. S. of pulmonary obstructive emphysema.

Syndromography

Clinic

The beginning is insidious. There are manifestations of intercurrent illnesses, progressive anorexia, weight loss, dyspnea, noisy breathing and cough.

Physical examination. Chest with deformity, increase in anteroposterior diameter, increase in intercostal spaces, elevation of the chest during breathing. Increased lung sound on percussion, prolongation of expiration, thick rales. Muffled heart sounds. Hepatomegaly. Finally cor-pulmonale with its physical signs. Clubbing fingers.

Paraclinical examinations

Chest radiology. Increased radiolucency, accentuation of the bronchovascular network. Findings of intercurrent pathologies. Reduction in lung vital capacity.

Blood. Low arterial oxygen saturation, carbon dioxide retention and polycythemia.

Syndromogenesis and etiology

The etiology may be hereditary in some cases and secondary to chronic lung diseases.

Literature

Fishman, A.P.; GM. Turino and E.H. Bergofsky: "The syndrome of alveolar hypoventilation. Amer. J. Med., 23:333-339.

Vargha, G; and P. Bruckner: Study of relationship between cavyti and obstructive ventilatory syndrome in tuberculosis". Amer. Review Respiratory Dis., 89:830-834, 1964.

KARTAGENER SYNDROME

Synonymy

S. of situs inversus, bronchitis and sinusitis. S. of Chandra- - Khetarpal.

Syndromography

Clinic

There is only one case reported in childhood. It occurs in adults of both sexes. It is characterized by cough, expectoration, and repeated pneumonia.

There is dyspnea, recurrent respiratory infections, palpitations, otitis media, nasal voice, progressive hearing loss.

Physical examination. Clinical evidence of situs inversus. Physical signs of bronchiectasis, mucopurulent nasal discharge, nasal polyposis, hearing loss.

Clubbing of fingers, diffuse fine crackles. Clinical findings of sinusitis.

Paraclinical examinations

Chest radiology. Pneumonic inflammatory infiltrates frequent.

Bronchography. Bronchiectasis. Dextrocardia.

Radiology of perinasal sinuses. Underdevelopment of the perinasal sinuses, sinusitis. Hyperemic nasal mucosa, polyposis.

Bacteriological sputum. Positive for pathogenic germs

Genos.

Immunoglobulin deficiency is common in childhood.

Syndromogenesis and etiology

It is a syndrome of genetic etiology and autosomal recessive inheritance whose basic defect is related to ciliary structures.

It has been suggested that the gene involved produces loss of control of normal levocardia, so that control of the position of the heart occurs at random, which explains the non-concordance of situs inversus expected in homozygous individuals.

Electron microscope observations have shown that the ciliated respiratory epithelium and spermatozoa show an absence of dyncin arms (protein structures that form temporary cross-bridges between adjacent ciliary filaments, responsible for generating movement in the cilia and the spermatozoon).

Taking into account that the cilium is composed of 100 different polypeptides, it is estimated that there is a high possibility that genetic heterogeneity exists in this syndrome.

On the other hand, it has been speculated that the embryonic cilium brings the heart to the left side around the fourth week in the mouse; if this is true in humans, this defect would be an example of a congenital malformation with functional abnormalities late in life.

Literature

Afzelius, B.A.: “A human syndrome caused by inmobile cilia.” Science,317-3 19, 1976.

Chandra, R.K., and S.K. Khetarpal: Levocardia with bronchiectasis and paranasal sinus abnormalities", IndianJ. Pediat.. 30:78-80, 1963.

Datta, P.: "Chandra's syndrome." Lancet, 2:1350-135 1, 1968.

Hartline, J.V., et al.: "Kartagener's syndrome in childhood." Am.J. Dis. Child., 121:349, 1971.

Hoeprich, P. de: Treatise on infectious diseases. t.1 Havana, Ediciones Revolucionarias, 1982. p.230.

Holmes, L.B.; J.B. Blennerhassett and K.E. Austen: “A reappraisal of Kartagener's syndrome." Amer. J. Med. Sci., 255:13-28, 1968.

Kartagener, M.: Zur Pathogenese der Bronkecktasien: Bronkectasien bei Situs viscerum inversus". Beitr. Klin. Tuberk., 83:489-501, 1933.

Kartagener, M., et al.: "Bronchiectasis with situs inversus." Arch. Pediatr., 79:193. 1962.

Moreno, A. and E.A. Murphy: "Inheritance of Kartagener syndrome." J. Med. Genet., 8:305-313, 1981.

MIDDLE LOBE SYNDROME

Synonymy

Brock-Graham S.

Syndromography

Clinic

It can be observed at any age. It begins with an acute febrile episode. Recurrent hemoptysis and pneumonitis, among the acute episodes there is chronic cough and fatigue. Chest pain.

Physical examination. Signs of bronchiectasis or chronic pulmonary suppuration.

Paraclinical examinations

Chest radiology. Image with radiopacity, right paracardial triangular appearance, atelectatic or inflammatory.

Bronchography. Obstruction of the bronchi that ventilate the middle lobe of the right lung.

Bronchoscopy. Obstruction of the same bronchial branches.

Syndromogenesis and etiology

Obstruction of the bronchi that ventilate the middle lobe of the right lung due to extrinsic compression (parabronchial nodes or tumors) or due to tumorous or obstructive endobronchial lesions.

Bibliography

Albo, R.J. and O.F. Grimes: "The middle lobe syndrome, a clinical study." Dis. Chest., 59:509-518, 1966.

Billig, D.M. et al.: "Middle lobe atelectasis in children. Clinical and bronchographic criteria in the selection of patients for surgery." Am. J. Dis. Child., 123:96, 1972.

Brock, R.C.; R.J. Cann and J.R. Dickinson: "Tuberculous mediastinal lymphadenitis in childhood; secondary effects on lungs." Guy's Hosp. Rep., 97:295-3 17, 1937.

Graham, E.A.; T.H. Burford and J.H. Mayerdisganma: “Middle lobe syndrome". Post. Grad. Med., 4:29-34, 1948.

LÖEFFLER SYNDROME

Synonymy

S. pulmonary infiltration with eosinophilia.

Syndromography

Clínica

It is characterized by weakness, anorexia, fever, cough, chest pain and dyspnea. Occasionally it may be asymptomatic.

Physical examination. Pleural rubs may be audible.

Pericardial effusion may also be detected. Prolonged and noisy expiration.

Paraclinical examinations

Chest radiology. Diffuse or nodular pulmonary infiltration, transient or transient.

Blood. Eosinophilia. Sputum: abundant eosinophils.

Syndromogenesis and etiology

Migration, through the lung, of larvae in patients parasitized by Ascaris lumbricoides, Necator americanus, Strongyloides stercoralis, Ancylostoma duodenalis and Fasciola hepatica.

Literature

Bahk, Y.W.: "Pulmonary paragonimiasis as a cause of Loeffler's syndrome." Radiology, 78:598, 1962.

Bailey, C.C., et al.: "Lymphosarcoma presenting as Loffler's syndrome", Br. Med. J., 1460, 1973.

Hardy, W., et al.: "The hypereosinophilic syndrome." Ann. Intern.Med., 68:1220, 1968.

Loeffler, W.: "Zur Differential-diagnose der Lungeninfiltrierungen: Uber fluchtige Succedan-infiltrate (mit Eosinophilie)". Beitraege Klin. Tuberculose, 79:338-367, 1932.

PULMONARY ALVEOLAR MICROLITHIASIS SYNDROME

Syndromography

Clinic

It affects all races in a familial way. It is more common in women, although sometimes there is no distinction between the sexes. The onset is insidious and is characterized by moderate dyspnea and ultimately the clinical picture of chronic cor pulmonale.

Physical examination. Cyanosis, edema in lower limbs.

Congestive hepatomegaly. Rales at lung bases. Hypertrophy of the right chambers of the heart.

Paraclinical examinations

Blood. Moderate polycythemia. Pulmonary function tests: alveolar ventilatory insufficiency, arterial hypoxia, decreased lung volume.

Chest radiology. Congestive image of both lung fields. On other occasions, numerous diffuse calcifications are observed.

Syndromogenesis and etiology

There may be several etiological factors and idiopathic forms:

1. Family form with autosomal hereditary characteristics.
2. Exposure to dusts rich in calcium salts; This condition may result from unknown alterations in the nature of the alveolar membrane wall that predisposes to this calcification.

Literature

Harbitz., F.: “Extensive calcification of the lungs as a distinct disease." Arch. Int. Med., 21:139, 1918.

O'Neill, R.P; J.E. Cohn, and E.D. Pellegrino: Pulmonary alveolar microlithiasis, a family study". Ann. int. Med., 67:957-967, 1967.

MOUNIER-KUHN SYNDROME

Synonymy

S. of trachyectasia. S. of megatrachea. S. of tracheal diverticulosis. S. of tracheobroncheomegaly. S. of tracheobroncheopathic malacia. S. of idiopathic megatrachea with tracheomalacia and

Syndromography

Clínica

Recurrent lower respiratory tract infections. Cough, dyspnea, mucopurulent-looking sputum.

Physical examination. Pulmonary signs of emphysema, crackles and subcrackles.

Paraclinical examinations

Radiology. Widening of the trachea and thick bronchi. Enlargement of the trachea with the Valsalva maneuver, saccular recesses, emphysema and pulmonary fibrosis. Presence of emphysematous

Syndromogenesis and etiology

Hereditary family, it has been reported in two cases of a family.

Literature

Aaby, G V., et al.: "Tracheobronchiomegaly." Ann. Thorac. Surg. 2:64, 1966.

Mounier-Kuhn, P.: "Dilatation of the tracheobronchiomegaly: "Constations radiographiques et bronchoscopiques". Lyon Med., 150: 106, 1932.

Zador, A.: "Tracheomegalie." Fortschr. Rontgenst., 119:498, 1973.

INTERSTITIAL DESCAMATIVE PNEUMONIC SYNDROME

Syndromography

Clinic

It is more common in adults. In 50% of cases it is preceded by a non-specific upper respiratory infection. Slow onset dyspnea, dry cough, weight loss, anorexia and asthenia.

Physical examination. Cyanosis in one in two adults. Drumstick fingers. Rest of the physical examination: there are no pathological data.

Paraclinical examinations

Chest radiology. Triangular opacity at the bases with the apex towards the hilum. Hilar lymphadenopathy may be present.

Electrocardiogram. Right ventricular hypertrophy.

Lung function. Hyperventilation, fall in arterial oxygen tension, elevated alveolar arterial oxygen gradient.

Lung biopsy. Massive proliferation of cell walls. Minimal interstitial inflammation, moderate thickening of the alveolar walls, absence of necrosis, normal bronchioles.

Syndromogenesis and etiology

The etiology is unknown.

Literature

Liebow, A.A.; A. Steer and J.G. Billingsley: "Desquamative interstitial pneumonia." Amer. J. Med., 39:369-404, 1965.

Schneider, R.M.; D.B. Nevius; H.Z. Brown: "Desquamative interstitial pneumonia in a four-year old child." New English J. Med., 277:1056-1058, 1967.

EOSINOPHILIC INTERSTITIAL PNEUMANTTIS SYNDROME

Synonymy

S. pulmonary infiltrative with eosinophilia, S. of the eosinophilic lung.

Syndromography

Clinic

The clinical findings are similar to those of Löeffler syndrome. Sometimes it presents recurrent episodes.

Physical examination. (See S. de Löeffler.)

Paraclinical examinations

Blood. Eosinophilia.

Spit. Eosinophilia.

Chest radiology. Infiltration of pulmonary inflammatory appearance that can be fleeting and transient. Other times images according to the etiology of the process.

Syndromogenesis and etiology

This syndrome can have different causes:

Parasitic: Ascaris, Trichuris, Endamoeba, Necator, Anchylostoma. Strongyloides, Echinococcus.

Infectious: tuberculosis, coccidiomycosis, brucellosis, viral and bacterial pneumonia, bronchiectasis.

Neoplastic: Hodgkin's disease, eosinophilic granuloma, eosinophilic leukemia.

Collagenosis: periarteritis, rheumatoid arthritis, rheumatic fever.

Allergic: asthma, hay fever, serum sickness, Dresslers syndrome, drugs.

Others: fat embolism, aspiration pneumonia, idiopathic familial eosinophilia with pneumonia.

Literature

Bailey, C.C. et al. : "Lymphosarcoma presenting as Loffler's syndrome." Br. Med. J., 1:460, 1973.

Citro, L.A. et al.: "Eosinophilic lung disease (or how to slice PILE.)". Am. J. Roentgenol., 117:787, 1973.

Hall, J.W.; M. Kozak and W.W. Spink: "Pulmonary infiltrates, pericarditis and eosinophilia." Amer. J. Med., 36:135-143, 1964.

Reeder, W.H. and B.E. Goorich: "Pulmonary infiltration with eosinophilia." (PIE. syndrome). Ann. Intern. Med., 36:1217- -1240, 1952.

PULMONARY ARTERY OBSTRUCTION SYNDROME DUE TO SCHISTOSOMIASIS

Synonymy

S. of cardiovascular schistosomiasis. S. of pulmonary obstruction in schistosomiasis. S. of pulmonary endarteritis. S. of protopulmonary schistosomiasis.

Syndromography

Clinic

It occurs in young adults who have been or resided in schistomiasis-endemic areas, and is characterized by progressive dyspnea and pain in the upper abdomen.

Physical examination. Cyanosis, jugular engorgement. Edema in the lower limbs, few pulmonary signs in contrast to severe dyspnea. Hepatosplenomegaly. Heart: systodiastolic murmur.

Paraclinical examinations

Stool. The presence of the parasite can be demonstrated; it is more useful and frequent to find it in the biopsy of the rectal mucosa.

Blood. Moderate anemia.

Electrocardiogram. Heart failure pattern right.

Chest radiology. Right ventricular hypertrophy. Dilation of the pulmonary arteries.

Syndromogenesis and etiology

It is due to infestation by Schistosoma mansoni with obliterating arteriolitis lesions due to repeated embolisms caused by the eggs of the parasite, granulomas, arteritis with fibrinoid necrosis and hyaline thrombi.

Literature

Belelli, V.: Les oeufs de Bilharzia haematobia dans les poumons". Union med. egiz. Alessandria, 1:22-23. 1884-1885.

Macieira-Coelho, E., and C.S. Duarte: "The syndrome of portopulmonary schistosomiasis." Amer. J. Med., 43:944-950, 1967.

ONDINE SYNDROME

Synonymy

Hypothalamic S. of alveolar hypoventilation.

Syndromography

Clinic

This syndrome is manifested by drowsiness that disappears with adequate pulmonary ventilation, apathy, and transient symptoms of diabetes insipidus.

Physical examination. Obesity, the persistence of alveolar hypoventilation after weight loss makes possible the differential diagnosis with Pickwick syndrome. There is thickening of the retinal veins, increased pressure of the cerebrospinal fluid, which disappears with adequate pulmonary ventilation. Cardiomegaly.

Paraclinical examinations

Negative lung pressure. Normal.

Chest radiology. No pulmonary pathological findings.

Cerebral arteriography. Normal.

Electrocardiogram. No right ventricular hypertrophy.

Blood. Absence of polycythemia.

Syndromogenesis and etiology

The association with hypothalamic symptoms and the differential characteristics with other hypoventilation syndromes indicate the possibility of an inflammatory, degenerative, neoplastic or functional pathology at that level.

Literature

Fishman, L.S.; J.H. Samson and D.R. Sperling: "Primary alveolar hypoventilation syndrome (Ondine's curse)." Amer. J. Dis., Child., 1 10: 155-161, 1965.

Giraudoux, J.: Ondine'. In Four Plays, New York, Hill Wang Inc., V. 1., 1958, p. 253.

Severinghus, J.W. and R.A. Michel: "Ondine's curse. Failure of respiratory center automaticity while awake." Clin. Res., 10:122, 1962.

PADDED-DASH SYNDROME

Synonymy

S. laryngotracheal traumatic.

Syndromography

Clínica

It occurs among passengers who suffer traffic accidents. Seat belts can increase this syndrome. Respiratory distress due to airway obstruction. Aspiration of saliva, liquids or solids into the airways, hoarseness and pain when swallowing.

Physical examination. The neck may be normal. Subcutaneous emphysema. Cartilage damage in the neck. Lacerations of the craniofacial mass.

Paraclinical examinations

Radiology of the craniofacial mass, cervical spine. To detect fractures.

Laryngoscopy. To detect organic lesions or the presence of foreign bodies.

Syndromogenesis and etiology

Laryngotracheal trauma and injuries caused by collision and hyperextension of the neck against the instrument panel (blackboard) of the car or other vehicle.

Literature

Butler, R.M., and FH. Moser: "The padded dash syndrome: blunt trauma to the larynx and trachea." Laryngoscope, 78:1172- -1182, 1968.

PANCOAST SYNDROME

Synonymy

S. of Pancoast-Tobias-Ciuffini. S. optic-costovertebral. S. de Ciuffini-Hare.

Syndromography

Clínica

Chest pain, more localized to one of the shoulders. Paresthesias, paresis or weakness with lack of muscle strength in one arm or the other.

Physical examination. Muscle atrophy of the shoulders, arm and hand. Enophthalmia, eyelid ptosis and miosis (Horner syndrome).

Paraclinical examinations

Chest radiology. It may show erosion of the first ribs and sometimes of the vertebrae at that level, with opacity due to a tumor mass at the level of the apex of the lung.

Bronchoscopy. A tumor can be located in dobronchial.

Bronchography. Obstruction of the bronchial branches of the apical lobe.

Sputum cytology. By washing or by endoscopic sampling, it is positive for neoplastic cells.

Syndromogenesis and etiology

It is due to a tumor of the pulmonary apex that invades the pleural dome and damages the sympathetic branches at the base of the neck and the stellate ganglion.

Literature

Pancoast, H.K.: "Importance of careful roentgen-ray investigations of apical chest tumors." J.A.M.A., 83:1407-1411, 1924.

Pancoast, H.K.: "Superior pulmonary sulcus tumnor characterized by pain, Horner's syndrome, destruction of bone, and atrophy of hand muscles." J. A.M.A., 99:1391-1396, 1932.

PICKWICKIAN SYNDROME

Synonymy

S. of obesity-cardiopulmonary. S. of hyperinsulinism. S. of obesity with hypoventilation.

Syndromography

Clínica

It occurs in 10% of severely obese adults.

It is rare in obese children, if it occurs in these there may be mental retardation. There is drowsiness, apathy.

Physical examination. Obesity, decreased respiratory expansion, cyanosis, Cheyne-Stokes respiration with nocturnal onset. Signs of right heart failure.

Paraclinical examinations

Blood. Polycythemia, hypoxemia.

Lung function studies. Alveolar hypoventilation, carbon dioxide retention, decreased vital and total capacity, decreased expiratory reserve volume.

Electrocardiogram. Right axial deviation, electrical changes suggestive of anterior face ischemia.

Radiology. Cardiomegaly with normal or increased pulmonary vascularization.

Syndromogenesis and etiology

It is caused by great obesity that limits normal breathing. Ascites and edema can occur in the course of right heart failure that can accompany the chronic hypoxemia syndrome. Hyperinsulinism may be present, which has been considered part of this syndrome, in which cardiorespiratory symptoms are the most relevant. Autosomal recessive inheritance has been suggested.

Literature

Burwell, C.S.; E.D. Robin; R.D. Whaley, et al.: "Extreme obesity associated with alveolar hypoventilation- a Pickwickian syndrome." Amer. J. Med., 21:811-818, 1956.

Dickens, C.: The Posthumous Papers of the Pickwick Club (London Chapman and Hall, 1837).

Falsetti, HL.; JS. Hanson and B.S. Tabakin: "Obesity-hypoventilation syndrome in siblings." Am. Rev. Resp. Dis. 90:105-110, 1964.

Metzl, K.; P. Keligers; J. Kantor, et al.: The Pickwickian syndrome in a child, an extreme example of psychoneurotic obesity." Clin. Pediat., 8:49-53, 1969.

Suzuki, M.: "Pickwickian syndrome and endocardial fibroelastosis: A possible pathogenic correlation". Am. J. Med., 53:123, 1972.

PULMONARY ALVEOLAR PROTEINOSIS SYNDROME

Synonymy

S. de Rosen-Castleman-Liebow.

Syndromography

Clinic

It occurs at all ages, more common in adult males. Most cases are exposed to wood dust, irritating fumes or other substances. Insidious presentation. In half of the cases they are classified as febrile pneumonic processes. Progressive dysnchia. Productive cough. Abundant and thick, yellowish sputum, rarely hemoptoic.

Low fever, asthenia, chest pain and weight loss. There may be heart failure in very severe cases.

Physical examination. Physical signs are scarce. Rarely audible rales. In some cases clubbing fingers, cyanosis in very advanced cases.

Paraclinical examinations

Chest radiology. Increase in the reticular network of the lung in a diffuse or nodular, perihilar form. There are no calcifications or lymph nodules.

Blood. Light polycythemia. Normal or moderate leukocytosis neutrophilia.

Bacteriological sputum. Negative.

Lung biopsy. Lung alveoli filled with a PAS-positive substance due to lipid-rich protein material.

Immunological. Determination of IgA shows low levels in serum and secretions.

Syndromogenesis and etiology

This syndrome has been reported in families and a genetic etiology and transmission by autosomal recessive inheritance has been proposed.

The basic defect is unknown.

Literature

Colon, A.R., Jr. et al.: "Childhood pulmonary alveolar proteinosis (PAP): Report of a case and review of the literature." Am.J. Dis. Child.. 121:481, 1971.

Preger, L.: "Pulmonary alveolar proteinosis." Radiology, 92:1291, 1969.

Rosen, S.H.; B. Castleman and A.A. Liebow: "Pulmonary alveolar proteinosis." N. Engl. J. Med., 258:1123-1142, 1958.

Woldman, L.: "Cerebral complications of pulmonary alveolar proteinosis. " Lancet, 2:733-735, 1961.k

BLOCKED LUNG SYNDROME

Syndromography

Clinic

Asthmatic status that does not respond to: steroids, adrenaline, aminophylline, or treatments with intermittent application of pre-

positive sion, in patients who have excessively used nebulizations with isoproterenol.

Physical examination. Marked decrease in inspiratory thoracic expansion. Marked decrease in vesicular murmur. Wheezing rales greatly reduced in relation to acute crises, due to the blockage caused by mucomembranous secretions accumulated in the respiratory tract.

Paraclinical examinations

Chest radiology. Increase in intercostal spaces.

Signs of emphysema.

Blood. Hypoxemia. Increased carbon dioxide.

Syndromogenesis and etiology

It is due to the excessive use of isoproerenol nebulizations, which cause a great irritating effect on the mucosa of the respiratory tract with the consequent accumulation of secretions in the mucosal walls.

Literature

Death from asthma (Annotation). Lancet, 1:1412-1413, 1968.

Keighley, J.F.: *Iatrogenic asthma associated with adrenegic aerosols". Ann. Intern. Med., 65:985-995, 1966.

FARMER LUNG SYNDROME

Syndromography

Clínic

It occurs more frequently in rural men, between the months of October to May, exposed to fungi derived from stored vegetables or grains. It is characterized by cough, dyspnea, general malaise and fever, hours after exposure.

Physical examination. Cyanosis, diffuse rales in both lung fields.

Paraclinical examinations

Blood. Moderate leukocytosis with moderate eosinophilia.

Radiology. Fine nodular mottling 3-5 mm in diameter, sometimes pneumonitis.

Immunological studies in agar.

Syndromogenesis and etiology

Hypersensitivity to a glycopeptide antigen produced by different fungi (thermopolyspore or polyspore) of grains or other stored vegetables. They produce a granulomatous inflammatory reaction to a foreign body in the lung parenchyma with secondary bronchopneumonia. In chronic cases, secondary fibrosis and organized endobronchial exudates occur.

Literature

Campbell, J.M.: "Acute symptoms following work with hay." Brt. Med. J., 2: 1 143-1144, 1932.

Emanuel, D.A., FLJ. Wensel; C.I. Bowerman, and B.R. Lawton: "Farmer's lung." Amer. J. Med., 37: 392-401, 1964.

BREEDER'S PIGEON LUNG SYNDROME

Synonymy

Breeder's Bird Lung S. S. of the bird breeder.

Syndromography

Clínic

It occurs in people who care for pigeons. There is general malaise, fever, chills, dyspnea, cough, arthralgia. It begins 4-6 hours after exposure.

Physical examination. Crackling rales in both lung fields.

Paraclinical examinations

Blood. Eosinophilia is a rare finding. Presence of specific antibodies.

Chest radiology. Marked increase in the bronchovascular network. Presence of fine nodulations and the reticulum of the lung, a product of interstitial pneumonitis.

Syndromogenesis and etiology

Hypersensitivity reaction to pigeons.

Literature

Reed, C.E.; A. Sosman, and R.A. Barbee: "Pigeon-Breeder's lung: a newly observed intertitial pulmonary disease." J.A.M.A., 192:261-265, 1965.

Unger, J.D.; J.N. Fink, G.F. Unger: "Pigeon-Breeder's disease." Radiology, 90:683-687, 1968.

RETRACTED LUNG SYNDROME

Synonymy

S. of the shrunken lung.

Syndromography

Clínic

It begins after the fourth decade of life and is characterized by non-productive cough and dyspnea on exertion. These symptoms are progressive.

Physical examination. Decreased respiratory expansion.

Generalized alveolar hypoventilation with cracking in the bases. There may be symptoms and signs of underlying diseases or those associated with the syndrome (lupus erythematosus, collagen conditions).

Paraclinical examinations

Radiology. It is quite characteristic and presents elevation of both diaphragms. Bibasal lamellar atelectasis that are a consequence of markedly decreased diaphragmatic mobility.

These signs contrast with the absence of alterations in the lung parenchyma.

Respiratory functional tests. They demonstrate that there is an extrathoracic restriction that is manifested by a decrease in static lung volumes and vital capacity, a reduction in compliance and a decrease in maximum static respiratory pressures and transdiaphragmatic pressure.

Selective electromyography. Demonstrates greater fatigue of the diaphragm and intercostal muscles.

Syndromogenesis and etiology

It is observed in the course of collagen conditions, including systemic lupus erythematosus. It is considered due to respiratory muscle weakness, the greater degree of which would cause retracted lung syndrome.

Literature

Wiedermann, H.P. and R.A. Mattnay: "Pulmonary manifestations of the collagen vascular diseases." Clin. Chest Medicine, 10 (4):677-722, 1989.

TRANSPLANT LUNG SYNDROME

Syndromography

Clinic

It occurs in homotransplanted patients following a decrease in the steroid dose. There is fever, asthenia and cough.

Physical examination. Rales in the middle part and bases of both lungs.

Paraclinical examinations

Chest radiology. Diffuse inflammatory infiltrate in both hemithorax.

Lung function tests. Alveolocapillary block.

Sputum. Presence of fungi or viruses. Thickening of the alveolar septa.

Syndromogenesis and etiology

Immune disorders (viruses, fungi, infection).

The etiology is unknown.

Literature

Rifkind, D.. T.E. Starzl, T.L. Marchioro, et al.: "Transplantation pneumonia." J.A.M.A., 189:808-8 12, 1964.
Slapak, M., H.M. Lee, D.M. Hume,: "Transplant lung a new syndrome." Brit. Med. J., 1:80-84, 1968.

SMOKER'S RESPIRATORY SYNDROME

Synonymy

S. of smoking.

Syndromography

Clinic

It occurs in smokers of all ages. It is characterized by a chronic course with exacerbations, especially after increasing cigarette consumption. They present cough and expectoration. Dyspnea, precordial tightness, and chest pain are frequently present.

Physical examination. Chronic pharyngitis. Chronic bronchitis with dry rales.

Paraclinical examinations

Chest radiology. Increase in the bronchovascular network.

Hilar congestion.

Blood. Tendency towards polyglobulia.

Syndromogenesis and etiology

It is caused by smoking.

Literature

Fogg, A.H.: "Queries and minor notes: 'allergy to tobacco smoke'". J.A.M.A.. 144:810-811, 1950.
Waldbott, GL.: "Smoker's respiratory syndrome., a clinical entity." J.A.M.A.. 151:1398-1400, 1953.

SWYER-JAMES SYNDROME

Synonymy

S. of unilateral pulmonary emphysema. Unilateral translucent lung S. S. of functional hypoplasia of the pulmonary arteries.
S. of unilateral bronchiectasis without atelectasis syndrome.

Syndromography

Clinic

It occurs in children and adolescents. There are recurrent lung infections.

Physical examination. Decreased unilateral lung expansion, noisy breathing, fine crackles.

Paraclinical examinations

Chest radiology. Increase in transparency of a lung or lobe, decrease in the bronchovascular network and the hilar region.

Bronchoscopy. No bronchial obstruction.

Bronchography. Decreased peripheral filling.

Angiography. Decreased filling of the arteries

lungs.

Syndromogenesis and etiology

Acquired inflammatory damage, injury to the peripheral bronchi with emphysema and bronchiectasis, functional insufficiency of the pulmonary artery and its branches.

Congenital agenesis and hypoplasia of the pulmonary artery are generally accompanied by greater symptoms due to cardiovascular and pulmonary anomalies.

Literature

Figueroa-Casas, J.C. and D.E. Jenkins: "Unilateral hyperlucency of the lung (Swyer and James syndrome) case report with fourteen years' observation" , Amer. J. Med., 44:301-309, 1968.
Swyer, P.R.; G.C.W. James: "Case of unilateral pulmonary emphysema." Thorax, 8:133-136, 1953.

CORK WORKERS SYNDROME

Synonymy

S. of suberosis.

Syndromography

Clinic

It occurs in cork workers. The onset is insidious, with cough, weakness, weight loss, sudden fever, and

hacking cough.

Physical examination. Fine diffuse rales in both ears

mithorax.

Paraclinical examinations

Blood. Eosinophilia. Presence of serum precipitin against cork dust.

Chest radiology. Increased bronchovascular pattern, diffuse nodular infiltration, infiltration with a reticular appearance. In the acute phase, patchy opacities are observed.

Syndromogenesis and etiology

Hypersensitivity to cork dust.

Literature

Avila, R. and T.G. Villar: "Suberosis-respiratory disease in cork workers." Lancet, 1:620-621, 1968.

Emanuel, D.A.: B.R. Lawton and F.J. Wensel: "Maple-bark disease. pneumoconiosis due to coniosporum corticale". N. Engl. J. Med., 266:333-337, 1962.

WEINGARTEN SYNDROME

Synonymy

S. of tropical eosinophilia. S. of tropical eosinophilic asthma. S. of tropical pulmonary eosinophilia. S. of eosinophilic lung.

S. de Frimodt-Moller.

Syndromography

Clínic

It is seen in inhabitants of the countries of the near and far East.

It can appear in people who have traveled to Eastern countries.

Its clinical course is one of crisis and remission with respiratory symptoms of cough, dyspnea, fever and weight loss.

Spontaneous remissions and relapses.

It is a rare condition of filariasis, characterized by the presence of eosinophilia, a positive circulating antibody test for filaria and microfilaria in tissues, but not in blood, observed more frequently in India, Indonesia, Sri Lanka, Pakistan and Southeast Asia. Meyers and Kouwenaar syndrome is established when, in addition, there is a notable enlargement of the lymph nodes and spleen due to the granulomatous reaction and fibrosis that occurs as a consequence of the progression of the eosinophilic inflammatory reaction. Chronic cough and nocturnal bronchospasms are common in this syndrome, also caused by pulmonary infiltration. Meyers and Kouwenar syndrome is observed more frequently in children and Weingarten syndrome in young men. Both have very similar symptoms, predominating an increase in lymph nodes in the former and chronic cough, nocturnal bronchospasm and pulmonary miliary infiltrates in the latter.

Physical examination. On auscultation, rales are heard, mainly wheezing, bilateral, located in the middle and base of the lungs.

Paraclinical examinations

Radiology. Diffuse reticulonodular findings in both lungs. Hilar lymphadenopathy in many cases. Inflammatory infiltration in the middle part and base of both lungs. Radiological signs of bronchopneumonia and fibrosis in chronic cases.

Blood. Marked eosinophilia. Circulating antibody test for filaria, positive. Determination of microfilariae in blood, negative. Sputum with eosinophilia.

Syndromogenesis and etiology

It is considered that the condition is produced as a consequence of an atypical response of the host to infection by several species of filariae, including Wuchereria

Studies carried out in animal models suggest that microfilariae are eliminated from the peripheral circulation and trapped in different tissues by an IgG-dependent cellular effector mechanism.

Sometimes the cause is not found and it is described as tropical eosinophilia syndrome.

Literature

Ciferri, E: "Human pulmonary dirofilariasis in the United States. A critical review." Am. J. Trop. Med. Hyg., 31:302, 1982.

Chernin, E.: “Sir Patrick Manson's studies on the transmission and biology of filariasis.” Rev. Infect. Dis., 5:148, 1983.

Frimodt-Moller, C. et al.: "Pseudo-tuberculous condition associated with eosinophilia", Indian Med. Gaz. 75:607, 1940.

Ubwadia, F.E.: "Tropical eosinophilia: A correlation of clinical, histopathologic and lung function studies". Dis. Chest, 52:531, 1967.

Weingarten, B.J.: Tropical eosinophilia. Lancet, 1:103, 1943.

RELATED RESPIRATORY SYNDROMES

Extrinsic allergic alveolitis syndromes

Pigeon breeder.
Farmer's lung.
Workers with bagasse (Bagazosis).
Workers with cork (Cork Worker).

Respiratory distress syndromes

Aspiration of the contents of the amniotic sac and its complications: atelectasis and/or emphysema, pneumonitis, pneumothorax.

Respiratory distress simulating cyanotic congenital heart disease.

Hyaline membrane.
Transient tachypnea of the newborn.
Wilson-Mikity.

Thoracic outlet syndromes

Cervical rib.
Costoclavicular.
Naffaziger.
First thoracic rib.
Steinbrocker.
Wright.

Syndromes with hypoventilation

Alveolar-capillary block.
Hypoventilation.
Ventilatory obstructive.

Syndromes with pulmonary infiltration and eosinophilia

Idiopathic familial eosinophilia.
Tropical eosinophilia.
Loeffler.
S.P.IE.
Weingarten.

Abaresthesia. Loss of pressure sensitivity.

Abathiesthesia. Loss of notion of segmental attitudes.

Abduction. Movement that moves a limb, or a limb segment, away from the median axis of the body.

Ablation. Action of surgically separating part or all of an organ.

Acalculia. Inability to carry out simple arithmetic calculations.

Acanthosis Nigricans. Skin disease characterized by hyperpigmentation and papillomatosis of the flexion folds, especially the armpits and neck.

Akathisia. Fear of sitting and inability to stay seated. V. TASICINESIA, CATISOPHOBIA.

Achondroplasia. Cartilage development defect in the epiphysis of long bones, which produces a form of dwarfism; It is sometimes seen in rickets.

Acrocephaly. conical shape of cranial vault. Hypsocephaly, Oxycephaly, pyrgocephaly, turricephaly.

Acrocyanosis. Permanent purplish discoloration of the hands and feet, with slight pain, but without trophic alterations, probably due to endocrinosympathetic disorders.

Acromicria. Atrophy or smallness of the limbs.

Acropachyderma. V. PAQUIACRIA.

Acroparesthesia. Vasomotor neurosis characterized by nocturnal crises consisting of a sensation of dullness, tingling and stinging in the hands, with hyperesthesia and hyperalgesia of the fingers, cold and paleness or cyanosis of the hands.

Tinnitus. acoustic hallucination; subjective noises.

Adduction V. ADUCTION.

Adenoma. Epithelial tumor, generally benign, with a gland-like structure.

Chromophobe adenoma. Tumor of the pituitary gland whose cells are not easily stained with either acidic or basic dyes.

Adenopathy. Disease of the nodes, especially lymph nodes.

Adiadochokinesia. Powerlessness to stop a mnotor impulse and replace it with another diametrically opposite one. Suppression or decrease in the ability to quickly practice successive voluntary opposite movements.

Adynamia. Lack or loss of vital or normal force.

Adiposity. Fatness, obesity, excessive accumulation of fat, general or local.

Adipositas. Adiposity.

Adduction. Active or passive movement that brings a limb or other organ closer to the median plane of the body.

Aphasia. General term to designate the disorder, defect or loss of the power of spoken, written or memic expression, due to an injury to the brain centers.

Aphonia. Loss or decrease in voice due to local causes (vocal cords).

Ageusia (geusis: taste). Lack of sense of taste.

Visual agnosia. Loss of the ability to transform simple visual sensations into perceptions themselves, so the individual does not recognize people or objects.

Agraphia. Inability to express thoughts in writing due to a central injury.

Absolute agraphia: agraphia for simple letters.

Acoustic agraphia: inability to write from dictation.

Amnesiac agraphia: loss of memory of graphic signs in which the patient can copy a word, but is not able to write spontaneously or by dictation.

Musical agraphia: agraphia for musical signs.

Optical agraphia: inability to copy what is written.

Albinism. More or less complete congenital absence of pigment in the skin, hair and eyes.

Alkalosis. Excessive alkalinity of the body's fluids, increase in the alkaline reserve of the blood due to excessive entry of alkalines or insufficient elimination of them.

Alexia. (lexis: word). Form of aphasia in which it is impossible to read due to the loss of the cortical centers of the association between graphic signs and the corresponding concepts.

Alopecia. Baldness; natural or abnormal thinning of hair.

Hypnagogue hallucination. Mental error in the perception of the senses, not based on an objective reality.

Amaurosis. Blindness, especially that which occurs without apparent injury to the eye, due to disease of the optic nerve, retina, spinal cord or brain.

Amblyopia: (opos: eye). Darkening of vision due to imperfect sensitivity of the retina without organic damage to the eye.

Amenorrhea. Lack of menstruation.

Amnesia. Complete loss of memory representations at a time in the individual's life.

Amusia (amousia disharmony): Inability to produce or understand musical sounds.

Instrumental amusia: loss of the ability to play an instrument ment.

Vocal motor amusia: inability to intone.

Receptive or sensory amusia: inability to distinguish musical sounds.

Anabolism. Process by which nutritional materials are converted into living tissue; It represents the first phase of metabolism, constructive, as opposed to the last phase or catabolism, destructive.

Anacusia. Total deafness.

Analgesia. Abolition of pain sensitivity.

Anarthria. Inability to articulate words. Subcortical motor aphasia.

Anasarca. Infiltration of serosity into the cellular tissues of the body; general dropsy, generalized edema.

Androgens. Said to be substances that have a masculinizing action or cause the appearance of secondary sexual characteristics. Hormones that control the physiological state of the secondary sexual characteristics, characteristic of the male.

Anemia. Literally, lack of blood; clinically, a decrease in blood mass or some of its components, especially red blood cells or hemoglobin.

Hemolytic anemia. That due to the destruction of red blood cells.

Normocytic anemia. Anemia due to reduction in the number of red blood cells without alteration in size or hemoglobin content.

Anesthesia. Total or partial deprivation of sensitivity in general, especially tactile sensitivity, due to morbid alterations or artificially caused.

Aneuploid. Chromosomal number that is not an exact multiple of the aploid number.

Aneurysm. Bag made up of pathological and permanent dilation of the walls of an artery or vein, filled with circulating blood.

Angina. In general, inflammation of the isthmus of the fauces, more commonly localized inflammation in the tonsils or adjacent parts.

Angina pectoris. Condition characterized by paroxysmal pain in the chest, sometimes radiating to the left arm, with suffocation, constriction and a feeling of imminent death, due to spasm of the coronary arteries.

Angor pectoris. v.ANGINA PEST.

Anhidrosis. Lack or decrease in sweat.

Anisocytosis. Inequality in the size of cells, especially red blood cells.

Anisocoria. Pupil diameter inequality.

Anomaly. Irregularity, a state contrary to the natural order.

Anorexia. Lack of appetite.

Anosmia. Lack of sense of smell.

Anosognosia (nosos: disease; gnosis: knowledge). Ignorance of the existence of a disease or of a limb affected by paralysis. Ignorance of his paralysis in the left hemipleics.

Ankylosis. Abolition or limitation of the movements of a movable joint.

Anxiety. Anguish and restlessness that accompanies acute illnesses, particularly with a sensation of precordial constriction, and that prevents the patients from calming down.

Antibody. Specific substance in the blood produced as a reaction to the introduction of an antigen and exerts a specific antagonistic action on the substance by whose influence it has been formed.

Apathy. Lack of feeling or emotion; emotional indifference.

Apperception. Term introduced by Bleuler to describe the identification of a homogeneous group of sensations with previously acquired analogous complexes, together with all their connections.

Apperceptive. Relating to apperception.

Aplasia. Incomplete or defective development of a tissue or organ.

Apnea. Temporary suspension of the respiratory act.

Apraxia. Complete loss of the ability to perform coordinated movements for a specific purpose, without paralysis or ataxia.

Ideomotor apraxia. Improper use of objects due to disorder in the appreciation of the true use of things.

Akinesia. Akinesia. Lack, loss or cessation of movements.

Arachnoid. Delicate meningeal membrane, intermediate between the pia mater and the dura mater in the brain and spinal cord.

Arachnoiditis. Arachnitis. Inflammation of the arachnoid.

Spider. Arachnid of the order Arachneinos; There are numerous species, all of which have hooked chelicerae with poison glands.

Atherosclerosis. Arteriosclerosis.

Arterioles. Small arterial branch

Arteriosclerosis. Chronic vascular disease that includes various lesions of unknown etiology, characterized by regressive and proliferative alterations of the intima, with or without alteration of the media and in which a wide variety of deposits can be found, isolated or simultaneously, including lipids, calcium and phosphates, as well as intercellular substances, including elastic fibers, collagen, bone and blood elements.

Arteritis. Inflammation of the artery.

Arthrogryposis. Flexion or permanent contracture of a joint.

Ascites. Dropsy of the abdominal cavity: accumulation of fluids in the peritoneal cavity due to exudation or transudation.

Suffocation. Word formerly used to designate the arrest tion of the heart. Nowadays it is synonymous with stopping or lack of breathing.

Asynergy. Disorder of the ability to associate elementary movements in complex acts.

Asystole. incomplete or imperfect systole; heart failure to achieve complete systole.

Asthma. Disease characterized by attacks of expiratory dyspnea of variable duration, with cough, wheezing and a sensation of constriction due to spasm of the bronchi.

cardiac asthma. Dyspnea due to acute myocardial failure.

Asthenia. Lack or loss of strength.

Ataxia. Lack or irregularity of coordination, especially of muscle actions, without muscle weakness or spasm.

Atelectasis. Lack of distension or dilation of the lung parenchyma.

Athetosis (athetos: not fixed). Disorder characterized by continuous involuntary movements that are quite slow and extravagant. tes, mainly of fingers and hands, usually due to a lesion of the striatum.

Atlas. First cervical vertebra, which supports the head.

Atopy. Displacement, ectopia.

Atresia. Occlusion of a natural opening.

Atrophy. Decrease in the volume and weight of an organ.

Aura. particular sensation or phenomenon that precedes the attack of an illness or paroxysm, especially an epileptic attack: it can be of a motor, sensitive, sensory, vasomotor, secretory or psychic nature.

Absence. Memory loss or momentary interruption of thought, which manifests itself accidentally for various reasons.

Autosome. Ordinary chromosome, as distinguished from an allosome.

Axis. Second cervical vertebra.

Azotemia. Term generally applied to designate the presence of urea in the blood.

Bacteriouria or baciluria. Elimination of bacteria through urine.

Batesthesia. Deep sensitivity (conscious proprioceptive). Sense of segmental attitude.

Benign. That it is not evil; that it does not recur; It is especially applied to tumors and infections.

Blepharitis. Inflammation of the eyelids.

Blepharochalasis or blepharochalasia. Relaxation of the skin of the par- pado, due to atrophy of the intercellular tissue.

Goiter. Tumor of the thyroid body that produces a bulge in the front part of the neck.

Bradycardia. Pulse slowness.

Bradypnea. Slow breathing.

Braditeleokinesia. Phenomenon of motor incoordination, by which an action is suddenly stopped before accomplishing its objective and is then performed slowly and with difficulty.

Brachycephaly. Relating to or belonging to the arm and head.

Brachycephalic. Short head, flattened at the back, cephalic index greater than 80°.

Brachydactyly. Abnormal cutting of the fingers and toes.

Bronchiectasis. Pathological and permanent dilation of one or more bronchi, congenital or acquired due to chronic inflammation of the same bronchus. It is characterized by foul breathing and paroxysmal cough with abundant mucopurulent expectoration.

Bulimia. Great voracity or insatiable hunger.

Bursa. Bag; especially mucous pouch,

Camptodactyly. Permanent flexion of one or more fingers, of the auricular (little finger) especially in the first interphalangeal joint (Landouzy).

Cachexia. V. WASTE.

Carcinoid. Term created in 1888 by O. Lubarsch to describe a tumorous condition of the intestine, of a benign nature.
Masson demonstrated the existence of a proliferation of intranervous argentaffin cells of the neurocrine type, whose cytoplasmic granulations are capable of reducing silver salts, called, for that reason, argentaffin cells, from which the name "argentaffinomas" given to these tumors is taken.
Syndrome described by Thorson and other authors, characterized by the presence of tumors, generally benign, that produce excessive amounts of serotonin or 5-hydroxytryptamine.

Carcinoma. Cancer or malignant tumor, made up of polymorphic epithelial cells with a tendency to infiltrate nearby tissues and metastasize.

Carcinomatosis. State of carcinomatous condition or carcinomas spread throughout the body.

Black heart. V. YESTERDAY'S DISEASE.

Cardiomegaly. Cardiac hypertrophy.

Darling. Indicates relationship with core; example: karriorrhexis (fragmentation of the nucleus).

Catabolism. Destructive metabolism; contrary to anabolism;
passage of tissues from a high plane of complexity or specialization to a lower one.

Cataract. Opacity of the lens or its capsule.

Catisophobia. Akathisia, tasikinesia.

Causalgia. Neuralgia characterized by an intense local sensation of burning pain, which is usually observed as a result of incomplete sections of the nerves; located, generally, in the median or sciatic nerves.

Headache. Headache.

Headache. Cephalgia.

Blindness. Deprivation or loss of sight.

Cellulitis. Diffuse inflammation of the connective tissue, especially the subcutaneous cellular tissue.

Cyanosis. Blue or livid discoloration of the skin and mucous membranes, due to the absolute increase in reduced hemoglobin in the blood.

Sciatica. Painful inflammation of the sciatic nerve, usually a neuritis characterized by paresthesia of the thigh and leg, tenderness in the path of the nerve and sometimes muscle atrophy. The pain is consistent or subject to exacerbations.

The disease generally attacks adult individuals.

Cyclitis. Inflammation of the ciliary body.

Cycloplegia. Paralysis of the ciliary muscle and subsequently of accommodation.

Kyphoscoliosis. Combination of kyphosis and scoliosis.

Kyphosis. Abnormal anteroposterior curvature with dorsal prominence of the spine.

Cylindroid. False or mucous urine cylinder, of various origins.

nes.

Fissure. Cleft, canal or sulcus, especially any of the cerebral sulci.

Sylvian fissure. The one that separates the anterior and middle lobes of the brain, bifurcated into two branches that comprise Reil's insula.

Ischemic intermittent claudication. Numbness, weakness and painful stiffness of a lower limb after some time of walking and which disappear with rest of the limb, phenomena due to the obstruction of the arteries due to arteriosclerosis.

Click. Brief, sharp, metallic ringing noise, early systolic, in the aortic or pulmonary focus, perceived in cases of alteration of the vessels of the pedicle of the heart.

Clinodactyly. Permanent curvature or deviation of one or more fingers.

Clone. Irregular, discontinuous. Which is related to clonus.

Clonic movements are contractions that determine isolated and sudden jerks or merged into a sustained contraction. They are accompanied by thermal elevation and rapid exhaustion.

Clonus or clone. Series of rhythmic and involuntary contractions, determined in a muscle or muscle group by the sudden and passive extension of its tendons.

Coilonychia. Nail concavity status; spoon-shaped nail.

Colibacillosis. Infection caused by colibacillus.

Coloboma. Mutilation or defect, especially a congeal fissure

nite somewhere in the eye.

Coluria. Presence of bile in the urine; coloration of urine by bile.

Coma. State of deep drowsiness with abolition of consciousness, sensitivity and motility, which appears in the course of certain diseases or after serious trauma.

Chondrodysplasia. Delay and irregularity in cartilage formation; Olier's disease.

Chondromalacia. Abnormal softening of cartilage.

Collusion. Symptom of certain forms of alienation that

It consists of the ease of answers and the recitation of imaginary facts, soon forgotten by the person who devised them.

Confusion. Lack of order, entanglement.

Congenital. Born with the individual. Innate. that exists since

birth or before birth. Not acquired.

Conjunctivitis. Inflammation of the conjunctiva.

Coombs, proof of. Any of two laboratory tests that detect certain antigen-antibody reactions.

Coprolalia. Repetitive onomatomania of voices and phrases related to excrement, filth and obscenities. Exaggerated use of obscene words.

Korea. Convulsive nervous disease with involuntary and irregular clonic muscle contractions, associated with irritability and depression and mental disorders.

Coreiform. Similar to Korea.

Coryza. Catarrhal condition of the nasal mucosa, associated with mucous or mucopurulent discharge from the nasal orifices. It can be acute or chronic.

Corticocerebral. Relating to the cortex of the brain.

Coxalgia. Pain in the hip.

Coxa valga. Deformity of the neck of the femur that produces manifest external rotation of the lower limb.

Coxa vara. Downward curvature of the neck of the femur.

Craniopharyngioma. Congenital tumor of the pituitary gland, developed at the expense of the remains of the primitive pharyngohypophyseal tract.

Cryptorchidism. Absence of one or both testicles from the scrotum due to the detention of these organs in the abdomen or in the inguinal canal in their normal migration.

Cryptorchidism. Cryptorchidism.

Chronic. Prolonged for a long time; opposite of sharp.

Quadriplegia. Paralysis of all four limbs; tetraplegia.

Restiform body. Lateral cord of the medulla, which appears to be a continuation of the posterior cord of the medulla and extends to the cerebellum; inferior cerebellar peduncle.

subthalamic body. Hypothalamus.

Dactylitis. Inflammation of a finger.

Weakness. Lack or loss of strength, asthenia.

Decubitus. Attitude of the body in a state of rest on a more or less horizontal plane.

Hippocratic finger. Club finger or drumstick; thickening of the terminal phalanges of the fingers and curve of the nail towards the palmar region, which gives the end of the finger the appearance of a club; pneumohypertrophic osteoarthropathy.

Deletion. In genetics, loss of genetic material from a

chromosome.

Delirium. Disorder of the mental faculties, with or without alteration of the moral ones, manifested by incoherent speech, nervous excitement and insomnia.

Delirium tremens. Delirium with tremor and intense excitement accompanied by anxiety, mental disorders.

Depression. In psychiatry, pathological sadness, frustration, melancholy.

Dermatitis. Skin inflammation, dermatitis; inflammatory dermatosis.

Dermatoglyphia. Study of the superficial eminences of the skin of the feet and hands for identification purposes.

Deamination. Separation of the NH group from a compound amine.

Dehydration. Separation of water from a substance or compound

position.

Lameness. Inability.

Disorientation. State of mental confusion in which the subject loses notions of place and time.

Deuteropathy. Secondary disease developed under the influence of another.

Diaphysis. What separates two parts or is located between them.

Target cells. "Blank" cell characteristic of Mediterranean Cooley anemia.

Diaplasia. Reduction of a fracture or dislocation.

Diarrhea. Frequent, liquid and abundant bowel movements.

Diathesis. Term of indefinite meaning, synonym of many

sometimes of dyscrasia, crasis, temperament, constitution, habit, but whose dominant concept is that of an individual, congenital, hereditary predisposition to become ill with a certain group of ailments.

Diplegia. Paralysis affecting equal parts of each side of the body;

bilateral paralysis.

Diplopia. Double vision of objects due to coordination disorders of the ocular motor muscles.

Dysarthria. Moderate degree of anarthria. Inability to pronounce words in which consonants are repeated.

Dychondroplasia. Chondrodysplasia.

Dycrasia. Term from ancient medicine that indicates alteration in the composition of the humors, especially the blood;

bad constitution

Dysphagia. Difficult swallowing.

Dysphasia. Moderate degree of aphasia; language disorder that consists of the lack of coordination of words in their proper order.

Dysphonia. Phonation disorder, sometimes synonymous with hoarseness.

Dysgenesis. Disorder of the ability to procreate.

Dysgraphia. Alteration of written language with difficulty when choosing the letters that form words. It may be due to organic lesions (aphasia) or functional lesions (psychasthenia). Cramp of the clerks.

Dyslalia. Expression disorder with anomaly of the language organs.

Dyslexia. Moderate or transient alexia, in which the patient only reads the first words well and then, without visual disturbance or other alterations, gets tired, being able to continue after a more or less long interval.

Dysmamesia. Chewing difficulty.

Dysmetria. Incorrect appreciation of the distance in the movements or muscular acts of their extension.

Dysmimia. Mime or gestural language disorder. Inability or difficulty in imitation.

Dysmorphia. Dysmorphism. Deformity.

Dysmorphism. Defective form of an apparatus or organ of the economy.

Dyspnoea. Difficulty breathing.

Dysostosis. Defective ossification; defect in the normal ossification of cartilage.

Dysplasia. Developmental anomalies.

Dyskeratosis. Alteration of the keratinization of epidermal cells.

Dystaxia. Difficulty in directing voluntary movements. Partial ataxia.

Dysthermy. Low-intense and long-lasting hyperthermia, in which the clinical examination does not find any cause to explain it.

Dystocia. Difficult, painful or slow birth.

Dystonia. Alteration of the tone or tension of a tissue or organ.

Dystopia. Abnormal situation of an organ; ectopia, dislocation.

Dystrophy. Nutrition disorder and consecutive state.

adiposogenital dystrophy. Fröhlich syndrome.

Dolichocephaly. Which has a relatively long anteroposterior diameter of the head (or cephalic). Whose cephalic index ranges between 65 and 74.

Dolichostenomelia. Congenital deformity of the limbs.

Pain. Painful impression experienced by an organ or part and transmitted by sensory nerves to the brain.

Sickle cell. Sickle-shaped red blood cell, characteristic of stroke

nocythemia.

Eczema. Acute or chronic inflammatory condition of the skin, caused by various causes, and presenting erythema, edema, vesicles, exudation, scabs, lichenification and scales in the stages of its evolution.

Echolalia. Automatic repetition of the words that the patient hears.

Ecomimesia. Involuntary and impulsive imitation of gestures or movements seen in another individual or animal.

Ectoderm. External sheet of the blastoderm that forms the epidermis, sense organs and nervous system.

Ectomesoderm. Layer of cells derived from the ectoderm and not yet differentiated from the mesoderm.

Edema. Excessive accumulation of seroalbuminous fluid in cellular tissue due to various causes: decrease in plasma OSmotic pressure due to protein reduction; increased hydrostatic pressure in the capillaries due to heart failure;

increased permeability of capillary walls or obstruction of lymphatic pathways.

Lymphatic edema. Infiltration of fluid from the lymphatic vessels.

Electrocardiogram (ECG). Graphic plots of the electrical currents that produce a broken line, with rises and falls, corresponding to the activity of the atrial and ventricular myocardium.

Elorudodruphitis. Cat scratch.

Emaciation. Extreme wasting due to morbid cause.

Embolism. Sudden obstruction of a vessel, especially an artery, by a body dragged by the bloodstream.

Embolism. Embolism.

Empyema. Formation or spillage of pus in a preexisting cavity, especially in the pleura.

Dwarfism, Dwarf quality.

Enanthem. Rash on a mucosal surface especially of the mouth and pharynx.

Encephalitis. Inflammation of the brain.

Enchondroma. Tumor of cartilaginous tissue, which develops inside a bone.

Endocardium. Endothelial membrane that lines the interior of the heart chambers and valves (valvular endocardium).

Endophlebitis. Inflammation of the internal tunic of the veins.

Ayerza's disease. Erythema with chronic cyanosis, dyspnea, hepatosplenomegaly, bone marrow hyperplasia and sclerosis of the pulmonary artery.

Banti disease. Primitive splenomegaly with anemia and cirrhosis of the liver.

Emphysema. State of a tissue distended by gases, especially the presence of air in the subcutaneous or lung cellular tissue.

Pulmonary emphysema. Abnormal distension of the nPoaes alveoli, with loss of elasticity of the alveolar wall. Abnormal sinking of the eye; Opposite of exophthalmia.

Eosinopenia. Abnormal deficiency of eosinophil cells in the blood.

Epicanthus. Congenital anomaly in which the skin fold covers the inner corner and caruncle of the eye. Characteristic of the Mongolian race but not uncommon in children of other races in whom there is incomplete development of the nasal bones.

Epiphysis. End of a long bone attached to the body or shaft of the bone by cartilage during childhood, but later forming part of the same bone.

Epilepsy. Essentially chronic nervous disease, which presents itself by more or less frequent attacks, sometimes characterized (grand mal) by sudden loss of consciousness, tonic and clonic seizures and coma, and other times (petty mal) by vertiginous sensations or other equivalents.

Episcleritis. Inflammation of the cellular tissue between the sclera and the conjunctiva.

Epistaxis. Bleeding from the nasal passages.

Epithelioma. Tumor composed essentially of epithelial cells.

Ecchymosis. Extravasation of blood inside the tissues.

Skin coloration produced by the infiltration of blood into the subcutaneous cellular tissue; cardinal.

Erythema. Diffuse or patchy redness of the skin, produced by congestion of the capillaries, which disappears momentarily due to pressure.

Erythema marginatum. Erythema in which several groups of Spots meet and form sinuous lines that surround a pale center.

Erythremia. Global hyperplasia of the bone marrow with marked and persistent increase in blood volume and total erythrocyte mass, marked polycythemia (up to 12 million per cubic millimeter or more), erythrosis or erythrocyanosis, neurological and vasomotor disorders, and often splenomegaly.

Erythroblast. Colorless nucleated cell of the bone marrow, origin of the red blood cell, which includes microblasts, megaloblasts and normoblasts; They are not normally seen in the

blood.

Erythroblastopenia. Hypoplasia of the erythroblastic tissue of the bone marrow, almost always associated with alterations in the other sectors of spinal hemocytopoiesis. The pure form (also called aplastic anemia) is exceptional in pathology.

Congenital ichthyosiform erythroderma. Congenital condition similar to ichthyosis, with redness of the skin, which mainly attacks the flexor surfaces.

Scaphocephaly. Keel skull deformity, that is, elongated and elevated in an anteroposterior direction and flattened transversely.

Saltly.

Scleroderma. Condition characterized by induration of the skin and more or less complete atrophy of it.

Multiple sclerosis. Sclerosis of the brain or spinal cord or both, characterized by irregularly scattered plaques.

Scoliosis. Lateral deviation of the spine.

Scotoma. Dark spot, more or less extensive, immobile, that

It covers a portion of the visual field or object being viewed, resulting from the insensitivity of the corresponding part of the retina.

Slough. Mass of gangrenous tissue.

Spherophakia. Congenital and familial malformation due to the sudden arrest of the development of the lens, as a consequence

aplasia of Zinn's zonule. Its association with bradydactyly has been noted. The name Sohar syndrome is a morbid condition characterized by the association of progressive deafness, spherophakia and chronic renal failure.

which occurs with a hereditary and family character.

Sprain. Violent sprain or distension of a joint without dislocation, which can lead to rupture of a ligament or nearby muscle fibers.

Spasticity. Spastic or spasmodic quality.

Spina bifida. Congenital malformation consisting of a fissure of the spine, especially the lumbosacral region, due to lack of welding of one or more vertebral arches, and through which the meninges and, sometimes, the spinal cord with a small amount of cerebrospinal fluid herniate.

Spinocerebellar. Relating to the spinal cord and cerebellum.

Splenomegaly. Increase in volume or hypertrophy of the spleen.

Sprue. V. SPRUE.

Schistocytosis. Accumulation of schistocytes in the blood.

Schizophrenia. Bleuler's term for dementia praecox, which, according to this author's interpretation, would be a splitting or fissuring of psychic functions.

Steatorrhea. Presence of excess fat in the stool.

Stenosis. Congenital or accidental pathological narrowing of an orifice or duct.

Sterile. Barren, bearing no fruit.

Stymenus. Ulcer of the vulva with sclerosis and hypertrophy of various nature: tuberculous, cancerous, syphilitic, lymphogranulomatous, etc.

Stomatitis. Inflammation of the mucosa of the mouth.

Squint. Deviation of one of the eyes from its normal direction, so that the visual axes cannot be directed simultaneously to the same point; heterotopia.

Laryngeal stridor. Condition of newborns, characterized by respiratory cornage, attributed to a spasm of the glottis.

Stupor. State of partial unconsciousness with absence of movements and reaction to stimuli; It is seen in certain severe forms of typhoid fever and in some forms of melancholy and in catatonia.

Euphoria. Feeling of well-being, satisfaction, natural or provoked.

Eunuchoid. Said to be a man with physiologically inactive testicles; It is also said of the falsetto voice in men.

Eunuchoidism. Developmental anomaly due to deficiency of the endocrine glands that govern the sexual organs, sometimes with the presence of secondary female sexual characteristics.

Euthyroidism. Normal functioning state of the thyroid gland.

Exophthalmia or exophthalmos. Abnormal protrusion or projection of the eyeball.

Exotropia. Divergent strabismus, abnormal outward rotation of one or both eyes (bilateral external deviation of the eyeballs).

Facoma. Lens tumor.

Phacomatosis. Generic term used to group neurofibromatosis, tuberous sclerosis, and Von Hippel-Lindau and Sturge-Weber syndromes.

Falciform. In the shape of a sickle.

Pharyngectasia. Dilation of the pharynx.

Fatigue. Condition and sensation of the body parts after their exaggerated activity.

Feminization. Development of feminine characteristics.

Shwartzman phenomenon. Vasculitis after repeated intravenous injection of bacterial endotoxins to experimental animals.

Phenotype. Set of the manifest properties of an organism, whether or not they are hereditary.

Fibroelastic. Composed of fibrous and elastic tissue.

Fibrosis. fibroid degeneration.

Hay fever. Allergic condition that occurs annually as spring or summer approaches, and is characterized by conjunctivitis and nasal colds accompanied by asthmatic symptoms, caused by inhalation of pollen from certain plants.

Arteriovenous fistula. Abnormal communication between an artery and a vein, traumatic or congenital.

Phlebolito. Calculus or concretion in a vein.

Phlogogenic. Which produces inflammation.

Flutter. V. FLIPING.

Phoneme. Name proposed by Wernicke to designate verbal auditory hallucinations (threats, insults, etc.).

Foramen lacerum. Torn hole.

Foramen magnum. occipital foramen.

Photophobia. Abnormal intolerance for light, especially light caused by eye conditions.

Fremito. Shuddering or vibration, especially that which is perceptible by palpation.

Galactorrhea. Abundant or excessive secretion of milk.

Gargolism. Heredofamilial disease characterized by nervous alterations similar to those of familial amaurotic idiocy, achondroplastic defects, mental deficiency and defective vision due to deposits in the cornea; lipochondrodystrophy.

Gastrectomy. Partial or total excision of the stomach.

Gastroenteritis. Inflammation of the stomach and intestines.

Gynadrism. Partial pseudohermaphroditism in women; development of the clitoris and welding of the labia majora.

Gynecomastia. Excessive breast volume in men.

Glaucoma. Eye disease, so called because of the greenish color that the pupil takes, characterized by increased intraocular pressure, hardness of the eyeball, atrophy of the optic disc and blindness.

Hysterical balloon. Subjective sensation of a ball in the larynx and strangulation that is often observed in hysterism.

Glossitis. Inflammation of the tongue.

Glossoptosis. Tongue falling backwards.

Gonadotropin. Substance of pituitary origin that stimulates the gonads; gonadostimulin.

Granulocytopenia. Lack of granulocytes in the blood; agranulocytosis.

Hallux varus. Separation of the big toe from the other toes.

Hematemesis. Vomiting of blood, coming from the stomach.

Hematoma. Tumor due to blood accumulation.

Hematopoiesis. Hemopoiesis.

Hemianalgesia. Analgesia in one half of the body.

Hemianopsia. Loss of vision in half of one or both visual fields.

Hemiataxia. Ataxia or motor incoordination that affects one side of the body.

Hemichorea. Chorea that only affects one side of the body.

Hemiparesis. Paresis of one half of the body.

Hemiplegia. Paralysis on one side of the body.

Hemoconcentration. Decrease in plasma volume without modification of the number of blood cells, resulting in a relative increase in the number of them per cubic millimeter.

Hemopoiesis. Formation or production of blood, especially its cellular elements; hematopoiesis.

Hemoptysis. Expectoration of blood in greater or lesser quantities.

Hemorrhage. Expulsion of blood from inside the vessels, due to accidental or spontaneous rupture of these.

Hemosiderosis. Deposition of hemosiderin in tissues, especially in the liver; hemochromatosis.

Hepatolenticular. Relating to the liver and the lenticular nucleus.

Hepatorenal. Relating to the liver and kidneys.

Hermaphroditism. Existence in the same individual of both sexes or of some characteristics of each sex; double or dubious sex.

Heterochromia. Color diversity of a part, especially the difference in coloration of both irises or parts of the same iris.

Hyaline. Translucent, homogeneous albuminoid substance that normally exists in cartilage, vitreous body, thyroid colloid, Wharton's gelatin, etc., and pathologically as a product of amyloid degeneration.

Hyalinization. Process by which the hyaline substance is deposited.

Hydramnios. Dropsy of the amnion; excess amniotic fluid.

Hydrocephalus. Accumulation of fluid in the brain due to increase of its production in the choroid plexuses of the ventricles or due to a decrease in its absorption. The most common variety in childhood is always due to obstruction, which can lie in the Luschka and Magendie foramina.

Hydromyelia. Internal hydrorachis. Spina bifida with serosity in the sac.

Hydronephrosis. Distention of the pelvis and renal calyces due to the accumulation of urine in these organs due to occlusion of

ureter and that forms a more or less voluminous fluctuating tumor.

Hydrothorax. Serous, transudate effusion in the pleural cavity, without previous inflammation of the latter, symptomatic of heart disease, venous obstruction, blood disorders, etc.

Hydroureter. Abnormal distension of the ureter by urine or a watery fluid.

Hyperazotemia or hyperaxoemia. Excess non-protein nitrogen (N.P.N.) in the blood.

Hypercalcemia. Excessive increase in calcium in the blood.

Hyperkinesia or hyperkinesia. Abnormal increase in motor function.

Hypercorticalism. Hyperactive function of the cortex of the adrenal capsules.

Hyperemia or hyperemia. Accumulation, active or passive, of blood that forms in a part or organ.

Hyperesthesia. Increase or exaggeration of general or special sensitivity.

Hyperglycemia. Excess glucose in the blood.

Hyperhidrosis. General or localized excessive sweating.

Hyperostosis. Neoformation or diffuse or localized hypertrophy of a bone.

Hyperparathyroidism. Exaggeration of parathyroid functions, translated by bone decalcification, hypercalcemia and muscle hypotonia.

Hyperpathy. Exaggerated sensation of pain. Extreme sensitivity.

Hyperpituitarism. Exaggerated activity of the pituitary gland and consecutive morbid state, characterized by gigantism or acromegaly.

Hyperpnea. Exaggeration in the amplitude and depth of respiratory movements.

Hyperkeratosis. Hypertrophy of the cone layer of the skin, or any skin disease characterized by it; keratoma.

Hyperreflexia. Exaggeration of reflexes.

Hypersecretion. Copious or excessive secretion, especially gastric: hypercrinia.

Hypersomnia. Excessive, pathological sleep.

Hypertelorism. Excessive separation between two parts or organs. This term was introduced by Greig in 1924, to denote the wide and abnormal separation between the two eyes.

Arterial hypertension. Abnormal elevation of blood pressure.

Hiccup. Singultus; Sudden spasm of the diaphragm and glottis, with shaking of the thoracic and abdominal walls and a high-pitched inspiratory sound.

Hearing loss. Decreased hearing sensitivity.

Hypocalcemia. Calcium deficiency in the blood.

Hypokinesia. Motor weakness, decreased movement or functional activity in living organisms.

Hypochloremia. Chloride deficiency in the blood.

Hypochlorhydria. Decrease in the proportion of free or combined hydrochloric acid in gastric juice.

Hypocolia. Decreased bile secretion; oligocholy.

Hippocratism. Hippocratic system of treatment, based on the observation and imitation of the processes of nature.

Digital hippocratism. Proliferation of soft tissue around the terminal phalanges of the hands and feet, sometimes with bony changes.

Hypophosphatemia. Decrease in the amount of phosphates in the blood or urine, respectively.

Hypoglycemia. Decrease in the amount of glucose contained in the blood.

Hypogonadism. Hypogenitalism.

Hypomimia. Weakening of emotional mimicry.

Hypopion. Accumulation of pus in the anterior chamber of the eye.

Hypoplasia. Decrease in training or production activity; incomplete or defective development.

Hypoponatremia. Deficiency of sodium salts in the blood.

Hyporeflexia. Decrease or weakening of reflexes.

Hypospadias. Congenital opening of the urethra on the underside of the penis.

Hypostenuria. Decrease in the concentration power of the kidney that results in low urinary density.

Hypostesia or hypoesthesia. Decreased sensitivity.

Hypothalamus. Subthalamus, vertebral portion of the diencephalon, which includes the mammillary bodies, the tuber cinereum, optic chiasm, lamina terminalis and pituitary gland.

Hypothermia. Decrease or drop in body temperature below normal.

Hypothyroidism. Deficient activity of the thyroid gland and consecutive state.

Hypotonia. Decreased tension or tone, especially of the muscles; lower intraocular tension; hypotension.

Hirsutism. Hypertrichosis, especially in women.

Ichthyosis. Skin disease characterized by dryness and excessive formation of scales.

Ictus. Sudden loss of consciousness.

Idiosyncrasy. Habit or temperament peculiar to each individual, a drug, duo. Peculiar or personal susceptibility to food or any agent; allergy and anaphylaxis.

Incontinence. Involuntary emission of materials whose excretion is normally subject to the will and controlled by sphincters.

Heart attack. Portion of parenchyma suddenly deprived of circulation blood loss due to obstruction of the corresponding artery, and a set of morbid phenomena consecutive to this obstruction.

Insomnia. Lack of sleep: abnormal sleeplessness.

Intercurrent. That appears during the course of another disease and modifies it in some way.

Intermittent. That has more or less regular rest or pause periods.

Lucid interval. Period of mental clarity in cyclical and recurrent psychoses or madness and in certain cases of cerebral confusion with subdural hematoma, between the period between the trauma and the appearance of chronological signs.

Intradermal. Intracutaneous, endodermal.

Ipsilateral. Located or occurring on the same side; It refers to the symptoms that appear on the same side of the brain injury that causes them.

Iridocyclitis. Inflammation of the iris and ciliary body.

Iritis. Inflammation of the iris, characterized by the change in color of the organ, contraction of the pupil, painful tension, photophobia and congestion of the ciliary region.

Isoimmunization. Immunization of an animal with antigen from the same animal species, such as the development of an anti-Rh serum by transfusion of Rh-positive blood into an Rh-negative individual.

Isostenuria. State of renal failure in which urine is secreted with always the same specific weight, corresponding to that of blood plasma without proteins.

Ischemia. local anemia; arrest of arterial circulation in a part and consecutive state of this part.

Jargonaphasia. Variety of aphasia in which the patient inserts meaningless syllables and words into the language, making it incomprehensible.

Infant. Child in the breastfeeding period, whether natural or artificial.

Lafora body (Lafora bodies). They are intracellular inclusions basophilic, PAS positive cells located in the cells of the cerebral cortex, substantia nigra, thalamus, globus pallidus and dentate nucleus.

Lateropulsion. Involuntary tendency while walking to move to one side.

Latrodectus. In North America the species L. mactans or black widow spider; It is very poisonous and its bite produces serious symptoms and even death.

Medial lemniscus. Medium Reil tape.

Leukopenia. Reduction of the number of leukocytes in the blood below 5,000 per cubic millimeter; hypoleukocytosis.

Livedo reticularis. It appears as a cyanotic coloration of the skin, but not diffusely, but rather in a mottled or reticular form, like mesh, and it intensifies with the cold.

Logorrhea. Immoderate penchant for speaking profusely or at length; tumultus sermonis, delirium of the tongue.

Macroglossia. Increase or hypertrophy of the volume of the tongue.

Megalocephaly. Abnormally large brain development.

Megalomania. Delusion of grandeur. Irrational conviction of one's own wealth, fame or power.

Microphthalmia. Congenital abnormal smallness of the eyes.

Microglossia. Congenital decrease in the size of the tongue.

Micrognathia. Congenital abnormal smallness of the jaw.

Myelomeningocele. Spina bifida with herniation of the spinal cord and its meninges.

Myoglobulinuria. Presence of myoglobulin in the urine.

Mutismno. Voluntary inhibition of language.

Negativism. Vesatic opposition to external influences that attempt to act on the patient's will, which can lead to the production of an action completely contrary to that advised or prescribed.

Nystagmus. Involuntary movements of the eyeball characterized by slow or fast jerks in a horizontal, vertical, or rotary direction, due to clonic contractions of the extrinsic muscles of the eye.

Orthopnea. Severe dyspnea that forces the patient to stand or sit.

Acute hemorrhagic pancreatitis. Inflammation of the pancreas due to a necroticohemorrhagic process inside the gland.

Panniculitis. Inflammatory hyperplasia of the subcutaneous fibrous tissue with the formation of small, painful nodules that are difficult to perceive.

Panophthalmia. Purulent inflammation of the entire eye; phlegmon of the eyeball; deep purulent ophthalmia.

Papule. Small, solid, circumscribed eruptive elevation of the skin, which usually ends by peeling; It is one of the basic skin lesions.

Pachyacria. State of marked thickening of the skin of the limbs, mainly club fingers, and bone deformities;
pseudoacromegaly or Brugsch syndrome.

Pachyonychia. Thickening of the nails.

Paraphasia. Partial aphasia characterized by the use of words inadequate or improperly applied.

Literal paraphasia. Letter change.

Syllabic paraphasia. Change of a syllable.

Verbal paraphasia. Changing one word for another.

Paralexia. Disorder of the ability to read, characterized by the transposition of words and syllables in meaningless combinations.

Paralysis. Loss of sensation or movement, especially the latter, of a part of the body.

Shaking paralysis. Parkinson's disease; chronic condition of the nervous system characterized by muscle rigidity, tremor, weakness and slowness of movement, hypertonia, hyporeflexia.

Flaccid paralysis. Variety in which the affected muscles are soft and in a state of complete relaxation.

Psychic paralysis. Hysterical paralysis.

Paranoia. Mental disturbance; in particular, chronic progressive functional psychosis, characterized mainly by systematized, logically constructed delusions: persecution, grandiose, litigious or complaining, etc.

Paraplegia. Paralysis of both lower limbs. It may be flaccid or spastic. The first can be central or peripheral (neuritis), while the second always recognizes a central, brain spinal origin.

Paresis. Partial or incomplete paralysis.

Paresthesia. Abnormal, strange, hallucinatory, tactile, thermal, etc. sensation of the senses or general sensitivity.

Paronychia. Whitlow, especially the periungual.

Parotitis. Inflammation of the parotid glands, generally resulting from a general infective state, characterized by their swelling and suppuration, with severe general symptoms.

you see.

Pellagra. Clinical syndrome that affects the skin (dermatitis), digestive tract (glossitis, aquilia, constipation or diarrhea), blood (anemia) and nervous system (tremor, convulsions, mental disorders, etc.). There is usually a history of an inadequate diet, rich in fats and carbohydrates; Its coexistence with beriberi and riboflavin deficiency is frequent.

Pemphigus. Group of dermatoses whose main elemental lesion is a blister, which always arises on healthy skin. These blisters are the result of acantholysis or loss of attachment of the spinous cells of the Malpighian mucosa.

Pericarditis. Inflammation of the pericardium.

Periodontium. Periosteum of the dental socket or pericementum.

Periostitis. Inflammation, acute or chronic, of the periosteum.

Peritonitis. Acute or chronic inflammation of the peritoneum.

Pycnosis. Condensation, thickness, especially degeneration cell in which the protoplasm becomes denser and the size of the cell decreases.

Plasmodium. Genus of protozoan microorganisms, parasites of red blood cells, causal agents of various forms of malaria in humans.

Platybasia. Developmental deformity in which the floor or lower portion of the occipital appears pushed upward by the vertebral column; It is also called basilar deformity.

Pleochromia. Increased coloration; It is generally said of feces as a consequence of pleocolia.

Plethora. Excess blood or other humors in the body or in a part of it.

Poikilocyte. Term applied mainly to deformed erythrocytes (pear-shaped, racket-shaped, etc.) that appear as a consequence of blood destruction during severe burns or, more often, due to a pre-existing alteration in the circulating blood (Cooley's disease, for example).

Poikilocytosis. State characterized by the presence of poikilocytes in the blood.

Poikiloderma. Rare dermatosis of unknown etiology, characterized by telangiectasia, pigmentation and atrophy: among them vascular atrophic poikiloderma.

Pollakiuria. Increased frequency of urine emission, not exactly polyuria.

Polyadenia or polyadenitis. Simultaneous inflammation of many nodes.

Polyarthritis. Simultaneous inflammation of several joints.

Polycythemia. Increase in the number of red blood cells, the blood; hyperglobulin or polyglobulia.

Polydactyly. Existence of supernumerary fingers.

Polydipsia. Excessive thirst; hydromania; Anadipsia.

Polyphagy. Voracious or excessive hunger.

Polymicrogyria. Smallness or poor development of the gyri brain tions.

Poliosis. Premature appearance of gray hair; canicie

Polypnea. Breathing with short, close and shallow inspirations and expirations.

Polyserositis. Inflammation of various serous membranes.

Polyuria. Abundant secretion and emission of urine. Total increase in daily volume.

Postprandial. After meals.

Prostration. Extreme despondency or exhaustion.

Precordial. Located in front of the heart.

Presbyophrenia. State of mental weakness common in old age, characterized by loss of memory and sense of place and by confabulation.

Priapism. Abnormal erection of the penis without sexual desire ordinarily, symptomatic of an inflammatory condition of the urethra and bladder, or of a spinal cord injury.

Proctitis. Inflammation of the rectum.

Prodrome. Sign, symptom or precursor state that indicates the beginning or approach of a disease.

Progeria. Premature old age.

Prognathism. Remarkable projection of lower jaw.

Proptosis. Fall forward: prolapse.

Proteus. Genus of bacteriaceae with multiple forms in mobile rods, gram-negative.

Protrusion. Abnormal advancement of a part, tumor or organ, due to an increase in volume or a subsequent cause that pushes them.

Boss. Eminence or elevation; apophysis. In neurology it means annular protuberance.

Pruritus. Particular sensation that encourages scratching.

Psychology. Branch of biology, which deals with the mind and emotional faculties.

Psychic. Psychology.

Pterygion colli. Congenital bridle of connective tissue and fascia from the mastoid region to the clavicle.

Ptosis. Fall or prolapse of an organ or part.

PTT. Partial thromboplastin time.

Puerperium. Overbirth; period from birth to woman
This means that the genital organs and general condition return to the previous ordinary state of pregnancy.

Purple. Condition characterized by the formation of red spots on the skin, consisting of small subcutaneous blood extravasations; symptom of various diseases.

Allergic purple. That caused by foreign proteins (allergens).

Putamen. External and darker portion of the lenticular nucleus of the striatum.

Cheilosis. Affection of the labial angles, especially due to vitamin deficiency due to riboflavin deficiency.

Burn. Injury produced in tissues by heat in its various forms.

Keratitis. Inflammation of the cornea.

Keratoconus. Protrusion of the comea due to its weakening.

Keratomalacia. Softening of the cornea.

Keratosis. Dermatoses characterized by an anomaly of the keratinization of the integuments.

Cyst. Tumor formed by a closed sac, normal or accidental, with liquid content.

Mucous cyst. Retention cyst, which contains mucus.

Rhabdomyolysis. Destruction of striated muscle.

Radiculitis. Inflammation of the spinal roots from the cord to the origin of the spinal nerves. Its symptom is radiculalgia (sensation of pain, caused by radiculitis).

Rash. Skin rash that has the morphological characteristics of a well-characterized disease.

RAST, Technique. (Radio allergo sorbent tecnic). It is satisfactorily related to the bioquantification of specific IgE; It can be performed by skin testing or histamine release from peripheral blood leukocytes and is convenient for patients;
however, it requires defined allergens and complete standardization.

Recessive. That it is capable of giving way or remitting, that it tends to move away or disappear.

Recurrent. That goes back or towards its origin.

Endurance. Opposition to the action of a force.

Reticulocyte. Young erythrocyte or proerythrocyte, which is derived from the acidophilic erythroblast. It represents 0.5 to 1% of circulating red blood cells in peripheral blood.

Retiform. In the form of a rope.

Retinitis pigmentosa. Progressive sclerosis of the retina, associated with pigmentation and atrophy, characterized by the formation of pigment deposits and obliteration of the vessels, decreased visual field and hemeralopia.

Retropulsion. Tendency to fall or walk backwards, observed in certain conditions of the nervous system, such as paralysis agitans.

Rigidity. Inflexibility, stiffness, especially abnormal or morbid.

Rinolalia. Phonation disorders determined by modifications in the resonance of the nasal cavities.

Rhinorrhea. Profuse flow of nasal mucus; nasal hydrorrhea.

Sacralization. Exaggerated development of the transverse processes of the fifth lumbar vertebra and apparent fusion with the sacrum.

ST segment. Portion of the normally isoelectric electrocardiogram, which begins at the end of the electrical depolarization period and includes the repolarization period.

Septicemia. Morbid state due to the existence of pathogenic bacteria and their products in the blood.

Shock or crash. In general, any sudden and severe vital depression due to trauma, emotion or impression on the nervous system, characterized mainly by insufficiency of peripheral circulation, drop in blood pressure, restlessness, anxiety and sometimes unconsciousness.

Shunt. Derivation. Anastomosis.

Sialorrhea. Exaggerated flow of saliva; salivation, ptyalism.

Syphilism or syphilosis. General syphilitic condition.

Babinski sign. Abnormal extension, instead of flexion
normal, of the toes when the sole of the foot is excited;
characteristic of organic hemiplegia, due to a lesion of the pyramidal tract or the anterolateral portions of the cord.

Kussmaul sign. Filling of the jugular veins during inspiration, observed in mediastinumpericarditis and mediastinal tumors. Thirst for air in patients in diabetic coma. Weakening of the pulse on inspiration; paradoxical pulse.

Sympathectomy or sympathectomy. Surgical, medical, or chemical ablation of a part of the sympathetic nervous system.

Sympathoblastoma. Malignant sympathetic neuroblastoma.

Synchondrosis. Mediate union of bones by cartilage, for example, of the ribs and sternum, of the sacrum and the iliac bone, of two adjacent vertebrae, etc.

Syncope. Fainting, fainting, lipothymia, generally secondary to acute cerebral anemia. Sudden apparent or real death due to cardiac paralysis.

Vasovagal syncope. Syncope in people with an unstable vasomotor system: strong emotion or pressure from the vagus produces a drop in blood pressure and a slow pulse.

Syndactyly. Skin adhesion, congenital or accidental, of two or more fingers to each other.

Synostosis (synostotic, synostotic0). Union of adjacent bones by means of bone matter; welding of bones, of the skull in particular.

Syringomyelia. Severe chronic condition of the spinal cord. Developmental defect that begins in the subependymal zone and consists of the proliferation and subsequent excavation of embryonic remains. The symptoms: a, sensitivity disorders, loss of pain and temperature sensations (thermoanesthesia), with preservation of the sense of touch (syringomyelic sensory dissociation); b, motor disorders; C, trophic skin disorders (glossy skin); d, vasomotor disorders, and e, spastic paralysis of the lower limbs and ataxia.

Reticuloendothelial system. Aschoff's term for a set of cellular elements of mesenchymal origin, spread throughout the organism, but mainly in the liver (Kupffer cells), spleen, lymphatics, bone marrow (clasmatocytes), with reticular and endothelial characteristics and to which hematopoietic, phagocytic, metabolic and immune functions and others are attributed.

Sprue. Megalocytic anemia, glossitis, diarrhea with steatorrhea, psychological disorders.

Stress. English word (effort, violence, tension) that designates the state of excessive tension as a result of an action, sudden or continuous, harmful to the organism. V. GENERAL ADAPTATION SYNDROME.

Subdiaphragmatic or subphrenic. Located or occurring below the diaphragm.

Black substance. Locus niger.

Smoking. Acute or chronic intoxication due to tobacco abuse; nicotinism or nicotism.

Tabes dorsalis. Syphilitic progressive locomotor ataxia.

Touch. Sense by which the palpable qualities of objects are known and whose organ lies in the skin.

Tachycardia. Acceleration of heartbeat.

Tachypnea. Rapid, shallow breathing.

Tasikinesia. Morbid tendency to move, to walk; akathisia or catisophobia.

Tegument. Wrapping, covering; skin or mucosa.

Telangiectasia. Abnormal dilation of small caliber capillaries, generalized or localized; simple angioma.

Tendinitis or tendonitis. Inflammation of a Teratoma tendon. Complex
embryonal tumor of multiple tissues that
It sometimes contains fetal remains, teeth, hair, etc., and is assumed to be due to the inclusion of an abortive fetus in another that develops normally.

Thermoanalgesia. Abolition of painful sensations produced by heat; thermoanesthesia.

Thesaurismosis. State resulting from the accumulation in the body of abnormally large quantities of normal or pathological substances (lipoids, phosphatides, glycogen, etc.).

Tetany. Intermittent neuromuscular hyperexcitability, related to a disturbance of electrolyte metabolism, especially calcium. Characterized by attacks of painful tonic contraction of the muscles.

Thrill. Shiver, rub. V. FREMITO.

Tibia. Largest and internal bone of the two that make up the skeleton of the leg.

Tinnitus. Subjective sensation of ringing or tinkling.

Thyrotoxicosis. Morbid state produced by excessive activity of the thyroid gland.

Torricephaly. Tower-shaped skull.

Stiff neck. Painful contraction of the sternocleidomastoid muscle.

Hemorrhagic toxicosis of the capillaries. Henoch's disease or purpura. V. S. DE HENOCH-SCHÖNLEIN.

Trigonocephaly. Dolichocephalus with a triangular head with an anterior vertex due to premature frontal synostosis.

Lockjaw. Tonic contraction of the chewing muscles, which produces forced occlusion of the mouth; symptom of tetanus and sometimes considered synonymous with it.

Thrombocytopenia. Plateletopenia or thrombopenia. Decrease in the number of blood platelets.

Thromboembolism. Embolism and complete occlusion of a vessel by a thrombus.

Thrombophlebitis. Phlebitis with introvascular clot formation, caused by a blood disorder.

Tuberculosis. Infectious, contagious and inoculable disease, caused by the Koch bacillus (Mycobacterium tuberculosis), characterized anatomically by the formation of tubercles, and by lesions and symptoms that vary depending on the location of the infection.

Ewing's tumor. Endothelioma of the bone marrow of long bones.

Sore. Breakout of continuity with loss of substance due to a necrotic process.

Uremia. Term applied to designate the set of toxic accidents caused by the accumulation, in the blood, of substances that the kidney normally eliminates.

Urticaria. Skin condition characterized by the sudden eruption of slightly raised plaques or wheals, of variable shape and size, accompanied by intense itching.

Uvea. The mediated vascular layer of the eye, which is divided from back to front into the choroid, ciliary body, and iris.

Uveitis. Inflammation of the uvea. V. ÚVEA.

Vasodilator. Which produces dilation of the vessels.

Vertigo. Alteration in the sense of balance, characterized by a sensation of instability and apparent rotational movement of the body or the objects present.

Virilism. Masculinity or masculinism; development of masculine characters in women.

Vitiligo. Leukoderma; skin condition characterized by the appearance of depigmented spots.

Vomit. Violent expulsion through the mouth of materials contained in the stomach.

Xanthelasma or xanthoma. Cholesterol deposit, especially that which affects the eyelids: xanthelasma palpebrarum.

Xanthochromia. Yellowing of the skin or other part. In the cerebrospinal fluid it is an indication of hemorrhage in the nervous centers.

Xanthoma. Skin condition characterized by the formation of more or less flat, yellow, slightly elevated plaques or nodules of various sizes, made up of cholesterol deposits.

Xerodermia. Skin dryness; asteatosis.

Xerostomia. Dry mouth due to lack of secretions; dry mouth, asialia.

GLOSSARY OF GENETICS

Balanced chromosome aberration. The existence of chromosomal abnormalities that do not imply an apparent loss of genetic information. They are characteristics of investments and translocations. Individuals who present this type of chromosomal aberration may face difficulties in the production of normal gametes and a history of reproductive failure.

Unbalanced chromosome aberration. This designation is used to refer to genomes with loss of information due to monosomies or with duplications of gametic chromosomes that generate trisomnias (aneuploidies) and also to refer to monosomies or partial trisomies as a result of structural defects.

Acentric. A chromosome fragment that has lost its centromere.

DNA. Polymer of nucleotides, in which the pentose is deoxyribose and the nitrogenous bases are adenine, thymine, guanine and cytosine. It constitutes the genetic material. It is found in the nucleus (nuclear DNA) and mitochondria (mtDNA).

Allele. One of the alternatives of a gene for a specific trait and that occupies the same locus. It is the result of mutations at that locus.

cAMP. Compound derived from ATP that acts as a second messenger in many information flows initiated by hormones.

Aneuploidy. This term refers to cells that have a karyotype with an inexact multiple of the haploid number of chromosomes:
2n-1 (monosomy), 2n+1 (trisomies), etc.

Apo. Prefix used to indicate the protein part in conjugated proteins. V. FOR EXAMPLE THE APOB.

ApoB. One of the proteins that is part of low-density lipoproteins (LD). Two forms are known: B48 formed in the small intestine and B100 that is synthesized in the liver. The latter participates in the mechanisms involved in cholesterol homeostasis.

RNA. Polymer of nucleotides, in which the pentose is ribose and the nitrogenous bases are adenine, cluracil, guanine and cytokine. They are the biomolecules through which the genetic message contained in DNA is transferred.

transfer RNA (tRNA). Types of RNA that participate in protein synthesis. There is one for each amino acid and they contain a structure of triplets of nitrogenous bases that are complementary to the triplets or codons of the mRNA and are called anticodons. V. CUPÓN.

Messenger RNA (mRNA). RNA structure that is synthesized from a segment of DNA that contains a gene, the information of which is transcribed therein. The primary transcribed RNA undergoes a maturation process during which the introns are lost and the exons remain V. INTRON AND EXON, ribosomal RNA (rRNA). Types of RNA that are found as part of ribosomes, cytoplasmic structures involved in protein synthesis.

Autosome. Any of the nuclear chromosomes excluding the sex chromosomes.

G bands. Technique used for longitudinal differentiation of the chromosome, using trypsin digestion before staining the slides containing the metaphases with Giemsa.

Karyotype. It is the result of the classification of chromosomes based on the photograph of a metaphase, taking into account the size and shape (according to the position of the centromere) of the 23 pairs of chromosomes in humans. It is the chromosomal constitution of an individual.

Centromere. Primary constriction of the chromosome that allows sister chromatids to hold together.

Codominant. Result of the simultaneous phenotypic expression of the two alleles in a heterozygous genotype.

Codon. A triplet of three bases in the DNA or mRNA molecule that represents a code for one of the 20 amino acids or that has specific information for the protein synthesis process.

Col2A1. Molecular structure of collagen type I.

Congenital. Present at birth and not necessarily genetic.

Chromatid. Chromosome structure that is observed in cell division and constitutes cytological evidence of DNA synthesis. Morphologically, they are observed as two parallel filaments joined by the centromere.

Sexual chromatin. Identification during the cellular interphase of the X or Y chromosomes. In the first case, it constitutes evidence of the late replication of one of the X chromosomes, known as the Barr body. It allows, using a simple technique, to obtain information on the presence or absence of them without the need to perform cell culture.

Ring chromosome. It is a chromosome that suffers breaks at both ends of the short and long arms and in the repair process both ends join together, generating an unbalanced chromosome aberration.

Deletion. Loss of a DNA sequence that can affect from a base to a chromosomal segment long enough for its absence to be detected by cytogenetic techniques.

Uniparental disomy. Genomic state in which the chromosomes representing a pair come from the same parent, so they can be of maternal or paternal origin. Taking into account, in addition, the moment of the meiotic error, they can be heterodisomies or isodisomies.

Dominant. Phenotype that is expressed from a mutated gene even when it occurs in a heterozygous state.

Exon. Region of a gene that is transcribed into mRNA.

Expressiveness. Grading of expression of a specific genetic defect fico. It is synonymous with clinical severity.

Phenotype. Conceptually it is the expression of a specific genotype. cific. This expression can be modified by the environment. Its study is not only morphological but also physiological, immunological, biochemical and even molecular. The level of your study brings you closer to knowing the genotype.

Gaps. Term used in cytogenetics to refer to a space observed along the chromatids of a chromosome. A break in continuity no larger than the width of the chromatid itself.

Gen. The unit of inheritance. In molecular terms it is a DNA sequence that is required for the production of a functional product.

Tumor suppressor gene. Normal gene whose protein product is involved in the negative control processes of cell proliferation. A mutation in this gene in double dose (autosomal recessive inheritance) or due to loss of heterozygosity (V. HETEROCYGOTIC), It is expressed by the absence or poor functioning of the tumor suppressor protein, allowing the development of a tumor, as occurs in retinoblastoma (RBl gene) or for the p53 protein (p53 gene).

Genome. It refers to all the DNA that contains the information genetics of a gamete, an individual, a species or a population.

Genotype. Genetic constitution for a locus that presents two or more allelic alternatives, always in combinations of two and that

They express a specific phenotype. The genotype can be homozygous, heterozygous or hemizygous.

Hemizygous. Genotype with a single representative of the genes of a pair of alleles, is used primarily to refer to the male genotype in X-linked inheritance, and to refer to the genotype as a result of partial or total monosomies of homologous chromosomes.

Mitochondrial inheritance. Special type of inheritance related to the transmission of mitochondria through the cytoplasm of the egg. Although men may be affected, they do not transmit this type of mutation since sperm do not contribute mitochondria to the fertilization phenomenon of the egg.

Multifactorial inheritance. Type of inheritance whose analysis of the family tree does not fit a Mendelian inheritance, but in which there is evidence of genetic participation of polygenes and environmental interaction. Congenital defects such as cleft lip or common chronic diseases such as diabetes, bronchial asthma, essential arterial hypertension are examples in which this type of inheritance is suggested.

heterozygous. Genotype that presents two alternative alleles at the same locus on homologous chromosomes.

obligate heterozygous. An individual who is clinically or phenotypically unaffected, but who, on the basis of segregation in the pedigree analysis, must have the mutated allele.

Heterodisomies. Regarding uniparental disomies, it consists of an error during meiosis I in which the two homologous chromosomes do not separate and go together to the same cell.

Allelic heterogeneity. Different alleles mutated for the same locus and that are expressed with a specific phenotype, providing it with different qualities. In the case that they express abnormality, their differences may correspond to different degrees of severity.

Clinical heterogeneity. Clinically different phenotypes produced by mutations of the same gene.

Genetic heterogeneity. Similar phenotypes expressed by different mutations and different pathogenetic mechanisms.

Influenced by sex. A trait whose gene does not have its locus on the X chromosome, however it is expressed with greater severity or frequency in the male or female sex.

Intron. A segment of DNA from a gene that, although transcribed into mRNA, is removed during the mRNA maturation process.

In the structure of a gene, introns are flanked by exons. V. ExON.

Investment. A rearrangement in a DNA sequence as a result of breakage and inverted repair of an affected segment. When the inverted segment is very extensive, it can affect the structure of the chromosome and be detected cytogenetically by banding techniques.

Isochromosome. An abnormal chromosome, the result of an error in the separation of sister chromatids, generates a chromosome with the same information by duplicating one of its arms and at the same time with loss of information or monosomy of the other arm.

Isodisomies. Regarding uniparental disomies, it consists of an error during meiosis II, in which the sister chromatids of a chromosome do not separate and travel together to the same cell.

kb (kilobase). A 1,000-base unit of a DNA or RNA sequence.

LDL. Low density lipoprotein. It is formed in plasma from very low density lipoproteins (VLDL) that have lost triacylglycerols in adipose tissue. They basically contain cholesterol that they transport to the extrahepatic tissues and participate in its homeostasis.

X-linked. Refers to genes located in the romosome X that segregate with this chromosome and that are therefore expressed in relation to sex when analyzing the family tree. When this occurs there is no transmission of the character in question from male to male.

Y-linked. Genes that have their locus on the Y chromosome.

Linkage. Genes located on the same chromosome very close to each other, segregating together to the same gamete, with a frequency much greater than expected, according to Mendel's second law. This condition is very useful in the process of mapping genes on chromosomes.

Limited to sex. A trait whose gene does not have its locus on the X chromosome, however, is forced to be expressed in the male or female sex due to the anatomical differences between the two.

Locus. The place a gene occupies on a chromosome.

Meiosis. Special cell division by which gametes (eggs and sperm) are produced, consists of two stages called meiosis I and meiosis I.

Monosomy. Phenomenon in which there is only one representative of the chromosomes of a pair that characterizes the diploid state of the organism. In humans the only viable total monosomy is the monosomy of the X chromosome.

Germinal mosaicism. Result of one or more mutations during the proliferation of germ cells, which give rise to two or more types of cells with different genotypes for a specific character from which cell lines are produced that can give rise to gametes affected and not affected by the mutation(s) referred to.

Mutation. Any permanent and hereditary change in the genomic or mitochondrial DNA sequence. Mutations can occur in germ or somatic cells, so their hereditary condition can be generational or in the process of tissue proliferation.

Dynamic mutation. Type of mutation that occurs in steps in gene areas with repeated triplet sequences, generating amplifications of the number of repetitions in ranges that define the normal allele, premutation and complete mutation and whose phenotypic effect explains the anticipation and severity of the genetic diseases they express. The most illustrative example is fragile X syndrome.

Germline mutation. It refers to mutations caused in the process of division of germ cells that will give rise to gametes, because all the cells, somatic and germ cells, of the individual resulting from the fertilization of a gamete affected by the mutation in question, will present the same genotype.

Somatic mutation. When the mutation occurs in a somatic cell, from which a cell line originates that expresses the abnormality caused by the mutation. Mutations of this type can have an onset as early as in the first divisions of the zygote.

Mutant. A gene that has been altered by a mutation. An individual carrying a mutated gene.

NAD. Oxidized form of NADH.

NADH. Enzymatic cofactor that participates in dehydrogenation reactions.

Nucleotide. A molecule composed of a nitrogenous base, a five-carbon sugar, and a phosphate group. A nucleic acid is a polymer of many nucleotides. V. DNA AND RNA.

Oncogene. The product of a mutation in a proto-oncogene that results in loss of its functions, causing uncontrolled cell divisions and therefore the development of a tumor. It is a dominant somatic mutation.

p. This letter identifies the chromosome arm above the centromere and since it is generally shorter, it was designated in French as petit (p).

Base pair (bp). A pair of complementary nucleotide bases

rios, as found in the double strand of DNA. It is used as a unit of measurement for the length of a DNA sequence.

PCR. Polymerase chain reaction, a technique that allows

You can amplify short DNA or RNA sequences more than 106 times the initial fragment, using small oligonucleotides flanking the area to be amplified.

Allows you to work with small amounts of DNA.

PEATC. Brainstem auditory evoked potential.

Penetrance. Concept that refers to the expression of a mutation that expresses a dominant phenotype. If this is expressed only in 80% of heterozygous people, for that mutation it is said that this mutation or mutated gene has a reduced penetrance, that is, 80% penetrance. Full penetrance is 100% and means that the mutated gene is always expressed.

Pericentric. around the centromere. Pericentric inversion (V. INVERSION), when there are two break points flanking the centromere and the resulting segment is repaired in an inverted manner.

Pleiotropy. It is the term used to refer to the effect, in more than one system, of a simple mutation.

Polygenic. Inheritance determined by several genes located at different loci and that have an additive effect on the phenotype they determine. The traits resulting from this type of inheritance have a greater predisposition to be modified by their relationship with the environment.

Polymorphism. It refers to the frequency with which alleles or alternatives of a locus appear in a population. An allele is considered polymorphic when its frequency is higher than expected due to new mutations, and a frequency of at least 0.01 has been determined, which means a frequency of 0.02 for heterozygotes.

Polyploidy. It refers to cells that have a genome (an exact multiple of the number of the species in question) greater than two. In humans 3(23) for a triploid genome, 4(23) for a tetraploid genome, etc.

Carrier. An individual heterozygous for a specific mutated allele. Individuals with heterozygous genotypes in autosomal recessive inheritances and in women with X-linked inheritances are carriers.

Probe or probe. It is an instrument used in genetic engineering to demonstrate the presence of the gene to be identified.
It consists of a simple sequence of radioactively labeled DNA or RNA that hybridizes with the sequence to be identified if both are complementary.

Prophase. The initial phase of cell division, during which chromosomes begin to become visible. In the prophase of the first meiosis, mechanisms also occur that contribute to genetic variation and allow the reduction of the number of chromosomes.

Promoter. DNA sequence located at the 5-prime end of the gene that determines the transcription initiation site, as well as the quantity and distribution of the mRNA in tissues.

Protein G. Family group of proteins that bind guanine nucleotides. If they are bound to GTP they are active, while if they are bound to GDP they are inactive. A family of them made up of three subunits participates in information transfer flows, generally in the next step to the membrane receptor.

Proto-oncogene. A normal gene involved in the positive control of cell proliferation.

q. This letter designates the chromosome arm below the centromere. It is the letter that follows the letter p in the alphabet.

Kinetochorus. A centromere structure to which achromatic fibers attach during cell division.

Receiver. Protein that participates in molecular information transfer flows. It binds to specific substances that reach the cell and triggers processes whose function is to adapt the cell to specific conditions. Receptors can be located on the plasma membrane or inside the cell.

Recessive. Term that refers to the expression of a character that is only expressed when in the genotype the mutated gene is found in a double dose (homozygous) or in a single dose (hemyzygous).

Recombination. The formation of new combinations of linked genes resulting from recombination between loci. Recombination takes place in the prophase of meiosis I and also mitosis of somatic cells.

RFLP, DNA fragments of different lengths with polymorphisms, which can be recognized with specific restriction enzymes.

Segregation. Term used to refer to the transmission of chromosomes or genes in gametes during meiosis.

Southern blot. A technique that allows the transfer of a DNA fragment, which has been separated by the electrophoresis process, in an agarose gel, to a nitrocellulose filter on which a specific DNA fragment can be detected by its hybridization to a radioactive "probe." V. "TRY".

Telomere. Ends of the chromosomes that have their own structure, which allows them to replicate.

Teratogenic. Agent that acts by interfering with the normal process of embryogenesis and that generates malformations, thus increasing their incidence. Teratogens can be biological, physical or chemical in nature or generated as a consequence of maternal endocrinometabolic defects. In addition, teratogens are a cause of reproductive failure.

Translation. Process by which the genetic information contained in mRNA is used to direct the synthesis of a protein.

Transcription. Process by which the information contained in DNA is transferred to RNA. Chemically it is the biosynthesis of RNA.

Translocation. Term used to describe the transfer of one chromosome segment to another. When this exchange occurs between acrocentric chromosomes, the translocation is reciprocal. When the translocation occurs at the centromeres of acrocentric chromosomes, it is called a Robertsonian translocation.

Trisomy. Phenomenon in which there are three representatives of a chromosome instead of the pair that characterizes the diploid state of the organism. In humans, the most common trisomy is chromosome 21.

VLDL. Very low-density lipoproteins that are formed in the liver and participate in the transport of triacylglycerols to adipose tissue. In the plasma they give rise to low-density lipoproteins (LDL).

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